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GENETIC TECHNOLOGY: PROMISES AND PROBLEMS

by

Mark S. Frankel

March 1973

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PREFACE

The purpose of this paper is to contribute to the discussion and evaluation of some new and emerging technologies of genetic medicine. The focus is upon the growing acquisition of new diagnostic capabilities, their consequent impact on screening and counseling for genetic disease, and the policy issues stemming from these capabilities.

It is becoming commonplace that modern technologies affect society in important and complex ways. They affect both our physical and psychic health and pervade our cultural and value systems, raising questions about the proper goals of society and about the proper means for pursuing those goals. They have an inescapable impact upon human life-styles, upon relationships between men, and upon man's understanding of himself. By its very nature technology promotes or retards what man considers valuable.

Such is the case with genetic technology. But this technology is unique: rather than merely acting upon man's environment, it operates directly on man himself. New genetic technology holds great promise for conquering disease and relieving human suffering. It is also providing man with the ability to unlock the most fundamental of life's processes, thus giving him the power to modify and perhaps control the quality and capacities of his species. Moreover, such actions may be irreversible. Genetic technology raises profound and complex questions for society and its policy-making processes: Who shall benefit from and

who shall pay for the use of this new technology? What are its probable effects on societal values and the distribution of political power? How will it affect our social and legal institutions? And on what basis will society allocate decisions to either personal conscience or public policy?

The new options created by genetic technology also raise profound questions about the appropriate priorities of our society. Faced with finite constraints on both money and manpower, decisions will have to be made regarding the support, use and control of genetic technology. These decisions are best made within the broader context of overall national goals, i.e. should a greater public investment be made in health or in military security?, as well as within the more limited scope of our health goals, i.e. should society continue to invest in more genetics research or should greater emphasis be given to distributing medical care on a more equitable basis? In addition, this new technology will increase the tensions between societal and individual values. Some just relationship between these values must be established in order to ensure the good of society while simultaneously protecting the rights of the individual. In each case, to choose one alternative or the other (or not to choose at all) implies an acceptance of certain uses of technology and not of others. Such choices are inherent in any social or political system and they are not neutral choices. They involve conscious judgments about what society considers valuable.

Conscious efforts to determine the desirability of a particular technology require a basic understanding of technological capability. This paper begins, therefore, with a "state-of-the-art" review which

is designed to provide a general and non-technical overview of current technological capability and the direction in which it is moving. The paper then explores some of the issues raised by the application of genetic technology. During the past several years, the mass media has given a good deal of emphasis to the more sensational applications of biomedical research, such as the creation of "test tube babies" and "genetic engineering."<sup>1</sup> Such promethean predictions have raised both hopes and fears among the public. While it is not the purpose of this paper to discuss the merits of such reporting, a major premise is that the sharpest challenges to our policy-making capability posed by genetic technology are already in view and pose more immediate policy concerns than do the more dramatic possibilities. The issues are indeed complex and the uncertainty which pervades them makes it clear that one needs to proceed cautiously in attempting to resolve them. Yet while the solutions and guidelines will take longer to construct, their propriety and efficacy will depend largely upon our ability to ask the proper questions and to identify the important issues. It is to these purposes that this paper is primarily directed.

The preparation of this paper was made easier because of the generous help of many people. Dr. Louis H. Mayo, Professor of Law and Director of the Program of Policy Studies in Science and Technology,

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<sup>1</sup>For example, a Durham Sun headline read, "Through Genetic Manipulation - Superhuman Race Said Possibility," April 8, 1967, p. 3; and a widely-read cover story of Time was titled, "The New Genetics: Man into Superman," April 19, 1971.

provided the initial idea and encouragement for the paper. John M. Logsdon, Director of George Washington University's Graduate Program in Science, Technology and Public Policy created a friendly environment in which I could work and provided helpful comments regarding a preliminary draft of the manuscript. Others who read earlier versions of the manuscript and whose comments were very helpful in preparing the final paper include John Fletcher, Cecil B. Jacobson and James R. Sorenson. I am also grateful to Dr. Jacobson for the opportunity to attend some of his genetic counseling sessions. My thanks also go to Robert Ehrhardt, Pat Faber and Martha Tappe and to Marcia Smith, whose patience and perseverance were instrumental in translating my thoughts onto paper. The preparation of this paper was partially supported by NSF Grant GS-34902. Of course, responsibility for the paper's content is solely mine.

In his study The Meaning of the Twentieth Century, Kenneth Boulding suggested that today we are "at the edge of a biological revolution, which may have results for mankind just as dramatic as the nuclear revolution of a generation ago."<sup>2</sup> While the nuclear age brought us powerful tools for benefiting mankind, it also brought overwhelming destruction and continues to generate feelings of fear and anxiety. Genetic technology offers a similar dilemma. Any attempt to resolve this dilemma in a consistent and humane manner will require a sober and anticipatory look at this "biological revolution."

M. S. F.  
March, 1973

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<sup>2</sup>  
(Harper and Row: New York, 1964), p. 7.



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## INTRODUCTION

Recent discoveries in medical genetics are permitting greater intervention into and manipulation of the genetic endowment of man. What was once accepted as a matter of fate is now becoming more susceptible to control by man. The rapid growth of various genetic technologies is providing an improved capability to diagnose, treat and eliminate numerous individual genetic qualities.

The improved capability to detect genetic defects in an unborn child now permits parents, in the case of some genetic diseases, to know if their child will be afflicted. If a genetic defect is diagnosed early enough in the pregnancy, parents can choose to terminate the pregnancy. On a much broader level, the ability to identify carriers of genetic defects by quick and simple medical tests permits large segments of the population to be screened and increases the potential for better control of genetic disease. Thus, couples who have not yet had their first child can learn whether they are at risk and, if they decide to conceive, can know whether their child is affected.

While this growing technological capability promises significant improvements in genetically-based health, it also raises complex questions regarding its application. For example, what defects are of such gravity as to justify intervention? What level of confidence in diagnostic capability is acceptable for making and implementing policy decisions? Should screening programs be voluntary or made compulsory? And by what criteria and by whom are these decisions to be made? Answers to these questions will be strongly influenced by and eventually reflect the interaction of prevailing values with increasing technological capability.

## GENETIC DISEASE: NATURE AND SCOPE

At least 1800 human diseases have been linked to a genetic cause<sup>1</sup>; new examples are being added to this list every year. In the United States, it is estimated that one of every 250 babies has a genetic defect which will result in mental retardation or physical disability<sup>2</sup> and that "at least 25 per cent of hospital beds and of all institutional places for the handicapped in this country are occupied by persons suffering some degree of genetic disease."<sup>3</sup> In addition to these affected persons, approximately 25 per cent of all conceptions are aborted naturally, and most of these can be related to an abnormal chromosome constitution or a lethal genotype.<sup>4</sup>

There are four types of genetic disease. The first of these includes single-gene disorders that may be either transmitted from one parent or both or might result from a new mutation. These diseases can be dominant, in which one of the parents is affected and each child has a 50 per cent chance of being affected (e.g. Huntington's chorea); recessive, in which both parents are normal

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<sup>1</sup>Victor A. McKusick, Mendelian Inheritance in Man, (The Johns Hopkins University Press: Baltimore, 1971). This figure includes those diseases for which the evidence for the particular mode of inheritance is judged to be incomplete, yet sufficiently strong to warrant inclusion.

<sup>2</sup>The NIH Record, Vol. 24, July 18, 1972, p. 8.

<sup>3</sup>Joshua Lederberg, Testimony on the 1971 Budget Appropriations for the Departments of Labor and Health, Education and Welfare, House Appropriations Subcommittee Hearings, 91st Congress, 2nd Session, Part 7, 1971, p. 915.

<sup>4</sup>Working Committee on Church and Society, "Three Reports From Church and Society," Study Encounter, Vol. 7, 1971 (World Council of Churches: Geneva), p. 5.

and each child has a 25 per cent chance of being affected, with 50 per cent of the children being heterozygous<sup>5</sup> and 25 per cent being homozygous<sup>6</sup> for the normal gene (e.g. sickle-cell anemia); or sex-linked,<sup>7</sup> in which a gene that produces a certain phenotypic trait is located on the X-chromosome (e.g. hemophilia). All genetically determined diseases related to an enzyme deficiency<sup>8</sup> are inherited by recessive or sex-linked mechanisms. A mutation involves a change in the genetic material which results in a new inherited variant. Most mutations are eliminated because they kill the cells in which they arise or their effects are countered by a preponderance of normal cells. Of those mutations that do surface in the individual, some are useful in helping him to adapt to his environment; the majority, however, are harmful and sometimes lethal to the individual in whom they are expressed.

The second type of genetic disease is exemplified by aberrations in the number and structure of chromosomes (e.g. Down's syndrome). In these instances, the abnormality is generally not transmitted, but is a result of a de novo event in the germ cells of the parents or in the initial division of the fertilized egg. It has been calculated that each year in the United States

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<sup>5</sup>An individual inheriting different genes from two parents is said to be heterozygous.

<sup>6</sup>An individual who inherits the same gene, defective or otherwise, from both parents is said to be homozygous.

<sup>7</sup>The 46 chromosomes of the human complement consist of 22 homologous pairs of autosomal chromosomes and a pair of sex chromosomes. The sex chromosomes are designated X and Y, the female having an XX constitution and the male one X chromosome and one Y chromosome.

<sup>8</sup>Victor A. McKusick has identified 92 disorders which are related to a specific enzyme. See "Human Genetics," Annual Review of Genetics, Vol. 4, 1970, pp. 9-11.

approximately 20,000 infants are born with a chromosome abnormality.<sup>9</sup>

Another category of genetic disease includes those caused by the interaction of multiple genes (e.g. diabetes). In these disorders, knowledge about the number of genes involved and how they interact is very limited. Yet the greatest proportion of those suffering from genetic disease are people with multigenic variety.

Finally, the fourth type of genetic disease is caused by incompatibility between fetus and mother, such as Rh hemolytic disease of the newborn.

When considered as a group, genetic disorders make up a highly visible and growing problem, resulting in significant individual and social burdens. Society's ability to cope with the illness which characterizes these diseases is, to a great extent, due to its increasing technological capability. What follows is a look at some of the more important techniques for the detection, treatment and prevention of genetic disease.

## GENETIC TECHNOLOGY: STATE-OF-THE-ART

### I. Intrauterine Diagnosis

The goal of intrauterine diagnosis is the detection of possible fetal abnormality in time to prevent its occurrence or to prevent disability from the initial defect by early treatment. There has been rapid progress in developing such techniques and while the number of disorders that can be diagnosed in utero is still relatively small, it is likely that the list will grow rapidly

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<sup>9</sup>H. A. Luks and F. H. Ruddle, "Chromosomal Abnormalities in the Human Population: Estimation of Rates Based on New Haven Newborn Study," Science, Vol. 169, July 31, 1970, p. 497.

as new and more sophisticated technologies are developed. Among the existing technologies are:

A. Amniocentesis: Abdominal amniocentesis involves the removal of approximately 10-20 ml. of fluid from the amniotic cavity by a syringe. Reliable cytogenetic and biochemical techniques have been developed to evaluate the intrauterine status of the fetus and some 40-50 genetic diseases can now be diagnosed by this method.<sup>10</sup> The cells are cultured and an individual's karyotype (his particular chromosome complement) is determined. The absence, duplication, or positional rearrangement of certain morphological features of the chromosomes, as compared with their typical topography, is visible evidence of chromosome mutation. In addition, the diagnosis of many inborn errors of metabolism is possible, based on demonstrating an abnormal level of enzyme activity in the cultured cells. It is likely that the increased availability and automation of assay procedures, including chromosome analysis, will expand the use of amniocentesis in the future.<sup>11</sup>

There are other considerations, however, which must be included in any assessment of this procedure. For instance, the most desirable time to perform amniocentesis - both in terms of the number of cells available for culture and of the minimal amount of risk to both the fetus and mother - is

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<sup>10</sup> Aubrey Milunsky, John W. Littlefield, Julian N. Kanfer, Edwin H. Kolodny, Vivian E. Shih and Leonard Atkins, "Prenatal Genetic Diagnosis," The New England Journal of Medicine, 283: 1370-1381, December 17, 1970.

<sup>11</sup> Theodore Friedmann, "Prenatal Diagnosis of Genetic Disease," Scientific American, Vol. 225, November 1971, p. 36; Also, Joan Lynn Arehart, "Prenatal Diagnosis: How Fast, How Far?," Science News, 100:44-45, July 17, 1971.

between the 17th and 20th post-menstrual week of the pregnancy.<sup>12</sup> Since the length of time required to acquire sufficient cells for biochemical analysis is four to six weeks,<sup>13</sup> the pregnancy will be in its 20-22nd week before a final diagnosis is made. If a therapeutic abortion is to follow, the long culture time raises several problems. Twenty to twenty-two weeks is past the time when most physicians would ideally prefer to perform an abortion. Furthermore, psychological and emotional problems (*infra*, p. 62) become more intense as the period of waiting increases.

There is also the problem of distinguishing between the affected homozygote and the heterozygote carrier. In the Lesch-Nyhan syndrome, a sex-linked disease characterized by mental retardation for which there is no treatment, the heterozygote and homozygote can be distinguished through prenatal diagnosis.<sup>14</sup> However, for most genetic disorders such a determination cannot be made.<sup>15</sup> When deciding whether or not to abort a fetus this technical problem raises important questions regarding genetic screening and counseling.

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<sup>12</sup> Interview with Cecil B. Jacobson, Chief of the Reproductive Genetics Unit, George Washington University Clinic, Washington, D.C., September 19, 1972. For a discussion of the problems involved in obtaining reliable cultures and minimizing the risk to both the fetus and mother, see Niels Hahnemann, "Possibility of Culturing Foetal Cells at Early States of Pregnancy," Clinical Genetics, 3: 286-93, 1972.

<sup>13</sup> M. M. Nelson and A. E. H. Emery, "Amniotic Fluid Cells: Prenatal Sex Prediction and Culture," British Medical Journal, Vol. 1, Feb. 28, 1970, p. 525.

<sup>14</sup> J. A. Boyle, K. O. Raivo, K. H. Astrin, J. D. Schulman, M. L. Graf, J. E. Seegmiller, and C. B. Jacobson, "Lesch-Nyhan Syndrome: Preventive Control by Prenatal Diagnosis," Science, Vol. 169, August 14, 1970, p. 688.

<sup>15</sup> Leonard Reisman and Adam Matheny, Genetics and Counseling in Medical Practice, (The C. V. Mosby Co.: St. Louis, 1969), p. 71.



Finally, one must also consider the risks involved. When amniocentesis is performed early (between 16-20 weeks) in pregnancy, the risks to the mother include bleeding, infection and blood group sensitization; those to the fetus include spontaneous abortion, fetal puncture and induced malformations.<sup>16</sup> Recent studies indicate that the occurrence of such risks during the second trimester of pregnancy is "less than 1 per cent."<sup>17</sup> At this time, however, very little is known about the long-term risks to the fetus.<sup>18</sup>

B. Ultrasound: High frequency sound is another method of prenatal diagnosis, with the sound either transmitted through a transducer or pictured on an oscilloscope screen. The advantage of this technique is that "all body tissues, except bone and air-filled lungs are relatively permeable to ultrasonic energy . . . This permits visualization, noninvasively, and with adequate resolution, of even the deepest structures in the body with ultrasound

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<sup>16</sup> Henry Nadler and Albert Gerbie, "Present Status of Amniocentesis in Intrauterine Diagnosis of Genetic Defects," Obstetrics and Gynecology, Vol. 38, November 1971, p. 794.

<sup>17</sup> Henry Nadler and Albert Gerbie, "Role of Amniocentesis in the Intrauterine Detection of Genetic Disorders," The New England Journal of Medicine, Vol. 282, March 12, 1970, p. 599. In what may be the longest on-going study of the risks involved in amniotic taps, Cecil B. Jacobson of The George Washington University reports that he has performed 580 taps with 100 per cent diagnostic accuracy. Only two pregnancies have been lost and both were confirmed as being independent of the amniotic tap. Paper presented at the American Association for the Advancement of Science annual meeting, Washington, D.C., December 28, 1972.

<sup>18</sup> In order to assess possible long-term risks, the National Institute of Child Health and Human Development of the National Institutes of Health is conducting an investigation in which patients not undergoing amniocentesis will be studied in parallel with those undergoing the procedure.

at sufficiently low intensity levels."<sup>19</sup> Moreover, the normal ultrasonic appearance of many fetal organs has been identified, making abnormalities of those organs susceptible to detection.<sup>20</sup> As a complement to amniocentesis, ultrasound can be used to determine the exact position of the placenta, which will permit greater accuracy and lessen the risk in withdrawing amniotic fluid.<sup>21</sup> A situation might also arise when diagnosis involves the presence of twins. The reliability of evaluation with amniocentesis in multiple pregnancies, however, is somewhat uncertain.<sup>22</sup> Ultrasound can be useful in these instances and, in fact, one study has reported a 99.75 per cent level of accuracy in diagnosing multiple pregnancies.<sup>23</sup>

The use of ultrasound is limited to some extent by the high cost of equipment and the scarcity of physicians trained to interpret the results. With respect to potential risks, a recent study has found some hazard with the application of this technique.<sup>24</sup> However, other investigators have been unable to reproduce these results even with longer exposure and higher

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<sup>19</sup> Padmakar P. Lele, "Application of Ultrasound in Medicine," The New England Journal of Medicine, Vol. 286, June 15, 1972, p. 1317.

<sup>20</sup> George Kossoff and William J. Garrett, "Ultrasonic Film Echography in Gynecology and Obstetrics," Obstetrics and Gynecology, Vol. 40, September 1970, p. 305.

<sup>21</sup> Joan Lynn Arehart, "Sounding Out the Womb," Science News, Vol. 100, December 25, 1971, p. 424.

<sup>22</sup> Nadler and Gerbie, op. cit., supra, n. 16 at 795.

<sup>23</sup> Ross E. Brown, "Doppler Ultrasound in Obstetrics," The Journal of the American Medical Association, Vol. 218, November 29, 1971, p. 1399.

<sup>24</sup> I. J. C. Macintosh and D. A. Davey, "Chromosome Aberrations Induced by an Ultrasonic Fetal Pulse Detected," British Medical Journal, 4:92-93, October 10, 1970. The investigators found evidence that chromosomal aberrations were induced in human lymphocytes following exposure to ultrasound. They employed intensities in the conventional Doppler diagnostic region (10-20 mWcm<sup>-2</sup>).

intensities.<sup>25</sup> The evidence seems to indicate that there is little risk associated with ultrasound, with a recent investigation concluding that "from all the evidence available, both experimental and clinical, it is conceded that the current diagnostic practices pose no short-term or long-term hazard to the patient or to the fetus."<sup>26</sup>

C. Fluorescent Staining: Whereas the usual karyotype exhibits only a silhouette of chromosomes, fluorescent analysis reveals numerous chromosome details. By using the chemical agent quinacrine or its derivative of quinacrine mustard, all the chromosomes in the human complement can be paired and fluorescent patterns distinguished, which can then be analyzed by a quantitative spectrofluorimetric method.<sup>27</sup> This staining method will increase the accuracy and reduce the time required for making prenatal sex determination,<sup>28</sup> though it will probably have to be used in conjunction with the traditional karyotype analysis.

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<sup>25</sup>E. Boyd, U. Abdulla, I. Donald, J.E.E. Flemming, A.J. Hall, M.A. Ferguson-Smith, British Medical Journal, 1:501, 1971.

<sup>26</sup>Lele, *op. cit.*, *supra*, n. 19 at 1317. This point is also supported by results from the following studies, all reported in the British Journal of Radiology, Vol. 45, May 1972: E.T. Coakley, J.S. Slade and J.M. Braeman, "Examination of Lymphocytes for Chromosome Aberrations After Ultrasonic Irradiation," pp. 328-332; C.R. Hill, G.P. Joshi and S.H. Revell, "A Search for Chromosome Damage Following Exposure of Chinese Hamster Cells to High Intensity, Pulsed Ultrasound," pp. 333-334; and P.L. Watts, A.J. Hall, and J.E.E. Flemming, "Ultrasound and Chromosome Damage," pp. 335-339.

<sup>27</sup>For a more detailed explanation of this technique, see D.A. Miller, P.W. Allerdice, O.J. Miller and W.R. Breg, "Quinacrine Fluorescence Patterns of Human D Group Chromosomes," Nature, 232:24-27, July 2, 1971. Another staining technique that does not require fluorescence microscopy has been described by Maximo E. Drets and Margery W. Shaw, "Specific Banding Patterns of Human Chromosomes," Proceedings of the National Academy of Sciences of the U.S.A., 68:2073-77, September 1971.

<sup>28</sup>Interview with Cecil B. Jacobson, *supra*, n. 12. See also A. Rook, L.Y. Hsu, M. Gertner, K. Hirschhorn, "Identification of Y and X Chromosomes in Amniotic Fluid Cells," Nature, 230:53, March 5, 1971.

Perhaps the most intriguing implication of the application of fluorescent staining is the observation of individual variation in the staining of the autosomes (a chromosome other than a sex chromosome).<sup>29</sup> If, as these results seem to indicate, the bands of particular chromosomes differ from one individual to another, then it might be possible to derive information from the chromosome directly that has heretofore only been inferred or inaccessible. For instance, in families with Huntington's chorea, a disease which does not manifest itself until after an individual has completed his reproductive years, it might permit the identification of those who are not gene carriers and thus free them to have children without risk. Of course, it will also allow the identification of those who are gene carriers; this raises sensitive problems for genetic counseling.<sup>30</sup> However, much more research into the technical aspects of this capability remains to be done before such identifications can be made. More imminently, however, this technique will make possible the observance of previously undetectable chromosomal translocations.<sup>31</sup>

D. Amnioscopy: This method permits direct visualization with a special conical endoscope of amniotic fluid. It has been used to facilitate intrauterine transfusion in fetuses with hemolytic disease and might be useful

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<sup>29</sup> P.L. Pearson, M. Bobrow, and C.G. Vosa, "Technique for Identifying Y Chromosome in Human Interphase Nuclei," Nature, Vol. 226, 1970, p. 80.

<sup>30</sup> A discussion of this possibility can be found in Lancet, 1: 275-276, February 6, 1971.

<sup>31</sup> Arthur D. Bloom, "Induced Chromosomal Aberrations: Biological and Clinical Significance," The Journal of Pediatrics, Vol. 81, July 1972, p. 3.

in recognizing gross fetal malformations or in obtaining fetal blood samples.<sup>32</sup>

E. Fetal Electrocardiography: This technique may be helpful for diagnosing potential genetic disorders, since changes in the heart rate "usually precede biochemical abnormalities rather than follow them."<sup>33</sup> It can also be employed to detect multiple pregnancies.

F. Fetal Sex Determination: Approximately 15 sex-linked genetic disorders can now be diagnosed prenatally.<sup>34</sup> However, at least 150 such disorders have been identified,<sup>35</sup> which means that the management of most of these diseases will depend upon the accurate determination of fetal sex. For example, in the case of hemophilia, where no distinction can yet be made between affected and normal males in utero, the disease could be prevented by prenatal sex determination if the mother were a heterozygote. Abortion of all male fetuses, whether affected or not, would guarantee that only normal daughters would be born.<sup>36</sup>

Amniocentesis provides a reliable method of determining fetal sex. Examination of cell nuclei for sex chromatin bodies (Barr bodies)<sup>37</sup> provides an inexpensive aid for diagnosing sex chromosome abnormalities. This method

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<sup>32</sup>Orlando J. Miller, "Intrauterine Diagnosis," in F. Clark Frazer and V.A. McKusick (eds.), Congenital Malformations: Proceedings of the Third International Conference, The Hague, The Netherlands, September 7-13, 1969, p. 351.

<sup>33</sup>Sheldon B. Korones, High-Risk Newborn Infants, (The C.V. Mosley Co.: St. Louis, 1972), p. 15.

<sup>34</sup>Interview with Cecil B. Jacobson, supra, n. 12.

<sup>35</sup>McKusick, op. cit., supra, n. 1 at ix.

<sup>36</sup>Aborting clinically normal male fetuses, of course, raises its own moral questions.

<sup>37</sup>The presence of the sex chromatin (Barr) body indicates a female. Its absence indicates a male.

is 100 per cent accurate, with two weeks required for developing the culture.<sup>38</sup> Fluorescent staining will reduce the diagnostic time to one day, but the level of accuracy may not be guaranteed.<sup>39</sup>

## II. Techniques for Genetic Screening

If prenatal diagnosis and selective abortion were combined with screening programs to identify heterozygous carriers, it would be possible to reduce the incidence of some recessive diseases. If screening procedures could identify couples in which both man and woman were carriers, the couple, after assessing the possibility of their having an affected child, could choose to abstain from having children or assume the risks involved in a pregnancy. In the latter case, the pregnancy could be monitored by amniocentesis and if the fetus were diagnosed as homozygous, an abortion could be performed.

Numerous large-scale screening programs for the detection of certain recessive disorders have begun to appear in the United States. For example, Tay-Sachs disease, a fatal cerebral degenerative disorder, can be detected in its heterozygous state by simple and rapid blood<sup>40</sup> and urine tests.<sup>41</sup> If necessary, amniocentesis can be used to monitor a pregnancy. The sickle-cell anemia trait can also be identified through a simple screening procedure which

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<sup>38</sup>Interview with Cecil B. Jacobson, supra, n. 12.

<sup>39</sup>A recent study has pointed out some of the technical problems involved in relying on fluorescent staining for sex determination. See A. Rook, et. al., op. cit., supra, n. 28.

<sup>40</sup>Jo Cohen, "New Aid for Tay-Sachs," Washington Post, July 16, 1972, pp. 9 and 14.

<sup>41</sup>R. Navon and B. Padeh, "Urinary Test for Identification of Tay-Sachs Genotypes," The Journal of Pediatrics, 80: 1026-20, June 1972.

requires only a small sample of blood.<sup>42</sup> However, there is not yet a proven method for detecting the sickle-cell homozygote in utero. Thus, unlike Tay-Sachs disease, abortion may not be a real alternative for carrier couples. The detection of heterozygous carriers is now possible for approximately 60 recessive genetic diseases<sup>43</sup> and it is likely that more funds will be made available and new screening programs implemented. Postnatal screening for sex anomalies can also be done quickly and accurately from buccal smear preparations (from the mouth) and the application of fluorescent staining to detect Barr Bodies should facilitate the screening of large populations.<sup>44</sup>

Screening programs which permit identification of the homozygous child after birth could, in the case of some disorders and if the diagnosis is made in time, permit proper treatment and care. The most frequently cited example is phenylketonuria (PKU), a disease caused by the absence of the enzyme responsible for the conversion of the essential amino acid, phenylalanine. Virtually all affected individuals experience mental retardation. If diagnosed early enough after birth, a special diet may result in the prevention of such retardation.<sup>45</sup> The incidence of PKU and/or hyperphenylalaninemia

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<sup>42</sup>The U.S. Congress recently passed the "National Sickle-Cell Anemia Control Act," PL 92-294, May 16, 1972, which provides for \$25 million for the fiscal year ending June 30, 1973, with \$40 million and \$50 million provided for the following two years. In addition, at least 12 states and the District of Columbia have enacted laws implementing some type of sickle-cell screening program.

<sup>43</sup>Bentley Glass, Letter to the Editor, Science, Vol. 173, July 9, 1971, p. 104.

<sup>44</sup>McKusick, op. cit., supra, n. 8 at 2.

<sup>45</sup>There is still much controversy over the reliability of existing diagnostic tests and the appropriateness of various treatments. (See infra, pp. 59-60.

has been estimated at 1 in 10,000 births<sup>46</sup>; at least 41 states have statutes which provide for the screening of the disease.<sup>47</sup> Finally, screening programs may serve a more immediate need for carriers of certain diseases and for those individuals whose genetic susceptibility to environmental agents is great. For instance, there is a growing body of evidence which shows that sickle-cell anemia in its heterozygous state is not the benign condition that it once was thought to be.<sup>48</sup> Thus, identification of the sickle-cell carrier (and perhaps of other disorders as well)<sup>49</sup> might make it possible to institute appropriate measures for therapy and care. Furthermore, some individuals possess a high

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<sup>46</sup> Benjamin White, "Case Finding of Phenylketonuria As a Public Health Approach," in J.A. Anderson and K.F. Swaiman (eds.), Phenylketonuria and Allied Metabolic Diseases, Proceedings of a Conference held in Washington, D.C., April 6-8, 1966. (U.S. Department of Health, Education, and Welfare, Children's Bureau, 1967), p. 152.

<sup>47</sup> Samuel P. Bessman and Judith P. Swazey, "Phenylketonuria: A Study of Biomedical Legislation," in E. Mendelsohn, J.P. Swazey and Irene Taviss (eds.), Human Aspects of Biomedical Innovation, (Harvard University Press: Massachusetts, 1971), p. 58.

<sup>48</sup> For evidence supporting this position see S. Brown, A. Merkow, M. Wiener and J. Khajezdeh, "Low Birth Weight in Babies Born to Mothers with Sickle-Cell Trait," The Journal of the American Medical Association, 221: 1405-08, September 18, 1972; and S.R. Jones, R.A. Binder and E.M. Donowho, "Sudden Death in Sickle-Cell Trait," The New England Journal of Medicine, 282 (Suppl. 6): 323, 1970.

<sup>49</sup> Thalassemia, or Cooley's Anemia, is another example. It is possible for carriers of the trait to be diagnosed as having mild iron deficiency anemia and given iron to correct the deficiency. Iron, however, is "contraindicated in thalassemia trait. It can be bad for the patient." Howard Pearson, quoted in B.J. Culliton, "Cooley's Anemia: Special Treatment for Another Ethnic Disease," Science, Vol. 178, November 10, 1972, p. 593.



genetic susceptibility to certain environmental agents.<sup>50</sup> More screening tests are becoming available which will forewarn these individuals so that they can reduce their exposure to such risks.

### III. Current Therapeutic Techniques

Much of the value of prenatal diagnosis and genetic screening programs will be contingent upon the availability of effective medical treatment. If such treatment is not available, however, the alternative of therapeutic abortion is usually considered.

A. Medical Treatment: As a result of recent scientific and medical advances, many hereditary disorders which previously had been considered incurable can now be prevented or controlled. Hemolytic disease of the fetus and newborn occurs once in 150-200 full-term pregnancies in the United States and each year an estimated 200,000 women face the possibility of having an affected child.<sup>51</sup> The disease involves the Rh incompatibility between mother and fetus and leads to severe anemia, brain damage and possible death of the fetus. The development of Rh immunoglobulin makes it possible to prevent the isoimmunization of an Rh negative woman to the Rh factor in her fetus. Thus, "the incidence of hemolytic disease of the fetus and newborn due to Rh isoimmunization can be practically eliminated once the present generation of sensitized women has passed the childbearing age."<sup>52</sup>

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<sup>50</sup>For some examples of genetic disorders susceptible to environmental agents, see V.E. Headings, "Genetic Susceptibility to Deleterious Effects of Environmental Factors," Medical Annals of the District of Columbia, Vol. 41, September 1972, p. 556.

<sup>51</sup>Jane S. Lin-Fu, Hemolytic Disease of the Fetus and Newborn Due to Rh Isoimmunization, (U.S. Department of Health, Education and Welfare, Maternal Child Health Service, 1969), p. 5.

<sup>52</sup>Ibid., p. 12.

The capacity to treat other genetic diseases is somewhat more limited. Common modes of treatment include surgery, diet and drug therapy, transplantation, and enzyme induction. The disorder called hereditary spherocytosis can be corrected by a surgical procedure, splenectomy, and plastic surgery has made less visible the effects of cleft lip malformations. Special diets permit the treatment of several metabolic disorders. For example, galactosemia is a disease in which the ingestion of milk sugar leads to stunted physical and mental growth and cataracts. These effects can be ameliorated by replacing milk with a synthetic formula containing cane sugar. In phenylketonuria, a reduced phenylalanine diet may substantially reduce the effects of the disease. Common drug therapy includes the injection of insulin to treat diabetes and of vitamin B<sub>12</sub> to treat pernicious anemia. Such dietary and drug therapy permits many people to live a much more tolerable life than would otherwise be possible, though there is actually no real "cure" of the basic genetic abnormality.

Transplantation may play an important role in treating some genetic disorders. In diseases such as thalassemia which affect the blood-forming organs, the transplantation of normal marrow may be an effective treatment. As with all transplants, however, there remains the problem of finding donors with genetically compatible tissue types.

The induction of certain enzymes by drugs can be useful in treating certain diseases. For example, the missing enzyme in hereditary jaundice can be stimulated by the introduction of phenobarbital. There have also been attempts to supply directly the deficient enzyme, but there are still several

technical problems associated with this procedure.<sup>53</sup>

While there has been substantial progress in developing different modes of therapy, most genetic diseases do not respond to such treatment. Even when disease management seems to be effective, there are concomitant problems to be considered. For instance, diabetics treated with insulin experience an increased incidence of vascular disorders compared to the normal population.<sup>54</sup> It has also been determined that the developing infant in a phenylketonuric woman is exposed to high concentrations of phenylalanine metabolites and, as a result, may experience mental and physical growth retardation and heart defects.<sup>55</sup> Thus, these constraints have encouraged attempts to develop a technology capable of treating genetic disease at a more basic level.

B. Gene Therapy: There are a number of promising techniques for treating genetic diseases at the genetic level. Although a detailed discussion of these techniques is beyond the scope of this paper, a brief overview follows.

If one could solve the problem of rejection, it might be possible to modify the genetic makeup of a tissue by cell replacement therapy. This involves taking cells or organs from a normal person and transplanting them

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<sup>53</sup>T. Friedmann and R. Roblin, "Gene Therapy for Human Genetic Disease," Science, Vol. 175, March 3, 1972, p. 950.

<sup>54</sup>S. Pell and C.A. D'Alonzo, "Factors Associated with Long-Term Survival of Diabetics," Journal of the American Medical Association, 214: 1833-1840, December 7, 1970.

<sup>55</sup>R.E. Stevenson and R.R. Howell, "Some Medical and Social Aspects of the Treatment for Genetic-Metabolic Diseases," The Annals of the American Academy of Political and Social Science, Vol. 399, January 1970, pp. 34-35.

into the person with a genetic disease. The normal cells would then either supplement the missing compound or completely take over the function of certain differentiated cells.<sup>56</sup>

An alternative to the above approach would be to supply the correct DNA to the defective gene. The most common method used to implement this approach is the introduction of new genes into mammalian cells. Current research indicates that animal viruses,<sup>57</sup> bacterial viruses,<sup>58</sup> and cell fusion techniques<sup>59</sup> are all capable of introducing new functional genes into mammalian cells. All of these techniques, however, are hampered by technical difficulties.<sup>60</sup> One problem involves directing the DNA material into the correct cell. There is also the failure of the added DNA to become permanently fixed with the existing DNA of the recipient cells.<sup>61</sup> Although the technology is still in the experimental stage, current research provides strong evidence to indicate that it will eventually become possible to transfer genes between,

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<sup>56</sup> Ernst Freese (ed.), Fogarty International Center Conference Report: The Prospects of Gene Therapy, (U.S. Department of Health, Education and Welfare, Publication No. (NIH) 72-61, May 1971), p. 2.

<sup>57</sup> P.K. Qasba and H.V. Aposhian, "DNA and Gene Therapy: Transfer of Mouse DNA to Human and Mouse Embryonic Cells by Polyoma Pseudovirions," Proceedings of the National Academy of Sciences of the U.S.A., 68:2345-2349, October 1971.

<sup>58</sup> Carl R. Merrill, Mark R. Geier and John C. Petricciani, "Bacterial Virus Gene Expression in Human Cells," Nature, 233:398-400, October 1972.

<sup>59</sup> A.G. Schwartz, P.R. Cook, and H. Harris, Nature New Biology, 230:5, 1971.

<sup>60</sup> For a discussion of these problems, see Friedmann and Roblin, op. cit., supra, n. 53 at 950-953.

<sup>61</sup> Recent experiments suggest that these problems may soon be resolved. See Merrill, et. al., op. cit., supra, n. 58.

and insert them into, mammalian cells.

Perhaps the greatest limitation on the use of gene therapy is the lack of knowledge regarding the fundamental genetic and regulatory processes of cells. According to one eminent biologist, "Far too little is known about possible dangers in this field to permit manipulations that might cause permanent distortion of generations of human beings . . ."<sup>62</sup> It might be helpful to ask what defects are of such gravity as to justify intervention? What might be the consequences of such intervention? For example, some genes may provide a selective advantage on heterozygous carriers. Heterozygotes for sickle-cell anemia are less prone to infection with malaria. Could the same be true with other genes? Could the cure be worse than the disease? There also "exists the possibility that groups of genes have been selected together, that is, they are co-adapted, and if one excises one of them, the effects may be greater than desired for the whole group of genes, some beneficial and some disadvantageous."<sup>63</sup> In light of present knowledge, there is a real problem in interpreting the effects of selection for or against particular genes.

Another difficulty applies to those diseases which are multigenic and are a result of combined hereditary and environmental conditions. Schizophrenia, manic-depressive illness and some forms of cancer are examples of diseases in

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<sup>62</sup>M.R. Pollock, "Molecular Genetics: Short-Term Application and Long-Term Possibilities," in Watson Fuller (ed.), The Biological Revolution, (Anchor Books: New York, 1972), p. 92.

<sup>63</sup>William J. Schull, "Population Genetics," in Arthur G. Steinberg (ed.), Conference on Genetic Disease Control, (Department of Health, Education and Welfare, National Institute of General Medical Services, February 1972), p. 33.

which genetic factors play an important part, but in which environmental forces are also at work. It is difficult to single out those genes responsible for these disorders. Even if they could be isolated and treated, environmental influences might be so strong that the effect of such treatment would be nullified. And finally, there is the fear that "uncontrolled tumor-like growth could easily be the consequence of introducing additional chromosomes or a host of viral genes."<sup>64</sup> At the present level of knowledge, then, a measure of caution might be the best "medicine" when applying gene therapy.<sup>65</sup>

C. Sex Predetermination:<sup>66</sup> The ability to predetermine sex will make possible the minimal perpetuation of sex-linked diseases, such as hemophilia and muscular dystrophy. If the X and Y fractions of semen could be separated, then insemination with either fraction would virtually assure control of the sex of offspring.

D. Abortion: For most genetic diseases there is no effective treatment or cure. Hence, if a defect is diagnosed in utero, an abortion offers a possible alternative.<sup>67</sup> The preferred technique for abortion prior to the

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<sup>64</sup>M.S. Fox and J.W. Littlefield, "Reservations Concerning Gene Therapy," Science, Vol. 173, July 16, 1971, p. 195.

<sup>65</sup>For some ethico-scientific criteria regarding the application of these techniques, see Friedmann and Roblin, op. cit., supra, n. 53 at 953-954.

<sup>66</sup>I have discussed both the technology and the potential consequences of predetermining sex elsewhere. See my monograph The Public Policy Dimensions of Artificial Insemination and Frozen-Semen Banking, (forthcoming, The Program of Policy Studies in Science and Technology, The George Washington University, Washington, D.C.)

<sup>67</sup>I recognize that abortion raises difficult social and ethical problems. Such problems, however, are beyond the scope of this paper and abortion is cited here simply to indicate that it is one additional alternative for responding to genetic disease.

first 12 weeks of pregnancy is dilation and suction evacuation. After 12 weeks gestation, an injection of a hypertonic saline solution is the most common method.<sup>68</sup> While there are possible complications associated with these methods,<sup>69</sup> there is a minimum of risk to the patient.

One factor which will help to determine whether or not abortion will be a real alternative for controlling genetic disease is the legal status of abortion. On January 22, 1973, the U.S. Supreme Court decided that the constitutional right to privacy is broad enough to limit the power of the states to regulate abortion decisions and the conditions under which they may be carried out, but that the right is not absolute. The Court divided pregnancy into three stages and balanced the woman's right to privacy against the State's interests in maternal health and in potential health. The Court ruled that

a woman had an absolute right during her first three months of pregnancy to decide whether to bear her child. Between the third and seventh months of pregnancy the states have the power to regulate the medical aspects of abortion. After the 26th or 27th weeks of pregnancy, the states may forbid all abortions except those essential to save the mother's life or health.<sup>70</sup>

The Court further facilitated a woman's efforts to obtain a medically approved abortion by striking down state residency laws for abortion services and eliminating procedural rules, such as requirements that abortions be approved

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<sup>68</sup>Selig Neubardt and Harold Schulman, Techniques of Abortion, (Little Brown & Co.: Boston, 1972).

<sup>69</sup>Edward A. Duffy, The Effect of Changes in the State Abortion Laws, (U.S. Department of Health, Education and Welfare, Maternal and Child Health Service, February 1971), pp. 8-9.

<sup>70</sup>Washington Post, January 23, 1973, pp. A1-2. Also included in the Court's opinion was the decision that a fetus is not a person under the Constitution and thus has no legal right to life.

by a special hospital committee, that have caused delays for women seeking official approval. As a result of the ruling, therefore, restrictive state anti-abortion laws have been declared unconstitutional.<sup>71</sup>

## APPLYING GENETIC TECHNOLOGY

The techniques reviewed in the previous section have improved considerably man's capability for controlling genetic disease. At the same time, however, their application raises new and complex issues. Questions concerning "when" and "how" genetic technology will be employed and who will be its "employer" are fraught with legal, social, ethical and political ramifications. And whether the answers to these questions will be framed within the context of long-range societal goals or the immediate amelioration of individual or family problems will have important consequences for developing policy. The remainder of this paper will explore some of these issues, emphasizing the variables which contribute to their complexity.

### I. Treating Genetic Disease

Two modes of therapy should be considered: current medical treatment and gene therapy. In the case of the former, most would agree that if an appropriate treatment is available it would be morally reprehensible not to provide it to all those in need. Modern medicine, guided by man's compassion for man and its commitment to the individual, makes no distinction between the sources and types of diseases or the individual "worth" of its patients.

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<sup>71</sup> Even the four states -- Alaska, Hawaii, New York and Washington -- that have permitted unrestricted abortions will probably have to alter their laws. These states have residency requirements which the Court struck down.



Prevailing professional and social mores "demand that all persons have recourse to all reasonable medical expertise . . ." <sup>72</sup> Providing proper care to all those in need, however, is not without its consequences.

A. Cost of treatment: One problem is the cost involved in providing the treatment. In the case of cystic fibrosis, the most common autosomal recessive defect in the white population, a recent study found that the costs were so high that "families who have been able to attain a moderate income . . . may be reduced to the poverty level by chronic health problems . . ." <sup>73</sup> The study also revealed the inadequacy of private health insurance, with the finding that 31 per cent of the children "were unprotected by medical insurance and only one-fourth had sufficiently comprehensive coverage to include outpatient expenses and medication." <sup>74</sup> Another example is hemophilia, for which the cost of treatment can be \$12,000 per year for each hemophiliac or approximately \$480 million for all Americans suffering from the disease. <sup>75</sup> Most American families would be unable to afford such treatment. Should the

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<sup>72</sup>Stevenson and Howell, op. cit., supra, n. 55 at 31.

<sup>73</sup>Audrey T. McCollum, "Cystic Fibrosis: Economic Impact Upon the Family," The American Journal of Public Health, Vol. 61, July 1971, p. 1340.

<sup>74</sup>Ibid. When insurance was available to families on an independent basis, the study found the cost of premiums to be as high as \$40-50 per month. Blue Cross has established a system under which families having a child with Cooley's Anemia can be reimbursed for transfusion costs even when their child is treated as an outpatient. The plan is being set up on a one-year trial basis and judgment regarding its effectiveness must be reserved until the end of that time. See B.J. Culliton, op. cit., supra, n. 49 at 591.

<sup>75</sup>K.M. Brinkhous, "Changing Prospects for Children With Hemophilia," Children, Vol. 17, November-December 1970, p. 222.

cost be absorbed by society? Can society then require that to receive such treatment a patient must fulfill certain conditions?

A recent case in Pennsylvania emphasizes the implications of the latter question.<sup>76</sup> A mother was initially informed by state officials that in order to receive treatment for her 12-year-old hemophiliac son she would have to go on welfare. The requirement was subsequently rescinded by the governor, who then told the family that they would have to recruit 36 donors a month and drive 100 miles to Philadelphia to donate blood in exchange for their son's treatment. The family, understandably perturbed, is now involved in negotiating a compromise with the State.<sup>77</sup> One must seriously question the intent of imposing such conditions on a family. Are there acceptable limits to such conditions? What criteria should be used for determining those limits? And with whom does that responsibility rest? A recent court suit demonstrates the challenge that lies ahead. Four young hemophiliacs have filed suit in federal court demanding that the government provide them and all other hemophiliacs in the country with the treatment necessary to allow them to bleed normally. The suit contends that the youths were denied the "equal protection" guaranteed by the 14th Amendment, citing the government maintenance programs for drug addicts.<sup>78</sup> Thus, the familiar problem of allocating resources becomes even more acute with the emergence of new genetic technology.

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<sup>76</sup>The following account is taken from articles appearing in the Washington Post, November 2, 1972, p. K3 and November 6, 1972, p. A20.

<sup>77</sup>As a result of this incident, it was recently announced that Pennsylvania had established a program to give free treatment of hemophiliacs. See the Washington Post, December 5, 1972, p. A4.

<sup>78</sup>Washington Post, October 11, 1972, p. A18.

B. Health Consequences of Treatment: Another unanticipated consequence associated with treating genetic disease can be illustrated by examining the treatment for PKU. As already noted (supra, p. 17), the fetus of a phenylketonuric woman is exposed to high concentrations of phenylalanine metabolites and, as a result, it may experience severe retardation and heart defects.<sup>79</sup> Thus, the medical science which enabled those mothers to lead relatively normal lives now threatens to deny their children the same benefits. Theoretically, correction of the metabolic disorder should prevent damage to the fetus. From a medical standpoint, however, reinstatement of the low phenylalanine diet poses two problems. First, "the health status of the phenylketonuric woman may not justify the difficulties involved in attempts to control diet."<sup>80</sup> And second, there is the possibility that reinstatement of the diet may "result in nutritional deficiency which may be as detrimental to the fetus as maternal phenylketonuria itself."<sup>81</sup> Under these circumstances, should the mother be encouraged not to conceive at all? If she insists on having a child, what is the physician's responsibility with regard to reinstating the special diet? Since the birth of such irreparably damaged children will result in life-long institutional care, does society have a right

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<sup>79</sup>According to R.R. Howell and R.B. Stevenson, "virtually every infant born to a woman who meets the criteria for classic phenylketonuria will have major defects, with growth retardation and microcephaly as well as other structural abnormalities." "The Offspring of Phenylketonuric Women," Social Biology, Vol. 18 (Supplement, 1971), p. S27.

<sup>80</sup>V. Elving Anderson, "Discussion: Maternal Effects in Genetic Disease," Ibid., p. S32.

<sup>81</sup>R.O. Fisch, D. Doeden, L.L. Lansky and J.A. Anderson, "Maternal Phenylketonuria: Detrimental Effects on Embryogenesis and Fetal Development," American Journal of Diseases of Children, Vol. 118, December 1969, p. 855.

to intervene in preventing such births? The consequences noted for PKU have been linked to other maternal disorders as well,<sup>82</sup> Thus increasing the proportions of the problem.

C. Treatment by Gene Therapy: Although the development and application of techniques for gene therapy are a number of years into the future, their potential impact warrants serious consideration at this time. The high degree of uncertainty and potential risks involved in using gene therapy clearly distinguish it from more conventional modes of therapy. Perhaps even more important, however, is that both the uncertainty and the risks will affect future offspring as well as the present generation. Friedmann and Roblin write that "For an acceptable genetic treatment of a human genetic defect, we would require that the gene therapy replace the functions of the defective gene segment without causing deleterious side effects in the treated individual or in his future offspring."<sup>83</sup> They conclude that "although the ethical problems posed by gene therapy are similar in principle to those posed by other experimental medical treatments, we feel that the irreversible and heritable nature of gene therapy means that the tolerable margin of risk and uncertainty in such therapy is reduced."<sup>84</sup> They support the need for continued research into the technology of gene therapy and propose ethico-scientific criteria for applying these techniques. However, they oppose using gene therapy in human patients at this time because of man's limited understanding of genetic processes and of how they might be affected by technological

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<sup>82</sup>Stevenson and Howell, op. cit., supra, n. 55 at 35.

<sup>83</sup>Friedmann and Roblin, op. cit., supra, n. 53 at 952.

<sup>84</sup>Ibid., p. 953.

should be prohibited or encouraged? What criteria or guidelines should be used for determining those conditions? And with whom do these responsibilities rest? These questions challenge both the scientific and ethical dimensions of the policy-making process.

D. Treatment and the Gene Pool: Perhaps the most frequently cited consequence of treating genetic disease is its impact on the human gene pool, i.e. the total genetic information possessed by the reproductive members of the population. The present gene pool is the result of 3 billion years of evolution and natural selection. Nature is successful in protecting the human species from detrimental genes because potential carriers either die prior to reproducing or reproduce less frequently than other hereditary types. The problem, as viewed by a growing number of people, is that medical advances have altered this situation by reducing the impact of natural selection. New treatment permits the survival and reproduction of persons with inheritable disorders who in earlier times would not have reproduced. As a result, the human gene pool experiences a higher frequency of many defective genes. For example, approximately 90 per cent of the children who formerly died from retinoblastoma - a malignant tumor of the eye - are now surviving because of advances in surgery and chemotherapy. Many of these children will be blind, but certainly able to reproduce and, consequently, to transmit the deleterious gene to their progeny.<sup>88</sup> Gene therapy which did not affect reproductive cells would produce similar results. Treated individuals would still pass the

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<sup>88</sup> Reisman and Matheny, op. cit., supra, n. 15 at 205.

intervention.<sup>85</sup> The tone of their presentation, however, suggests that gene therapy should and will become a useful method for medical treatment. Theologian Paul Ramsey, however, is less certain about the use of such therapy. He writes that "the unknown and unforeclosed risks to future generations may outweigh any benefit that might be secured for the individual patient. In a matter of such grave importance, 'no discernible risk' is not adequate protection. We need to know that there are no risks - a requirement which inheritable gene therapy is not apt to meet."<sup>86</sup> Ramsey is undoubtedly correct when he contends that complete knowledge regarding the possible risks of gene therapy is unlikely ever to be realized. Under such circumstances, then, gene therapy affecting future generations would be prohibited if Ramsey's criterion was observed. Ramsey believes that the choice is not simply between doing nothing about an inherited disease and correcting it by gene therapy. Alternative choices would include "having no children or fewer children. The treatment would be continence or not getting married or using three contraceptives at the same time or voluntary sterilization."<sup>87</sup> In light of the uncertainty and high risks involved in gene therapy, Ramsey finds more acceptable these other alternatives for "treating" genetic disease.

The two points of view outlined above converge at the crucial policy questions: Are there any conditions under which certain types of gene therapy

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<sup>85</sup> Ibid., p. 954.

<sup>86</sup> Paul Ramsey, "Genetic Therapy: A Theologian's Response," in Michael Hamilton (ed.), The New Genetics and the Future of Man, (Wm. B. Eerdmans Publishing Co.: Michigan, 1972), p. 169.

<sup>87</sup> Paul Ramsey, Fabricated Man, (Yale University Press: New Haven, 1970), p. 118.

defect on to their offspring, thus requiring more and more gene therapy.

What are the likely consequences if the genetic load is permitted to increase? According to some, the quality of the gene pool will continue to deteriorate and greater demands will be placed upon the community's medical services, since more people will be dependent on medical care and treatment. Ramsey foresees "some future generation [which] will begin to experience 20 percent genetic deaths."<sup>89</sup> And Bentley Glass draws the following scenerio:

to contemplate the man of tomorrow who must begin his day by adjusting his spectacles and his hearing aid, inserting his false teeth, taking an allergy injection in one arm and an insulin injection in another, and topping off his preparations for life by taking a tranquillizing pill, is none too pleasant.<sup>90</sup>

What is good for today's individual and his generation may be detrimental to future populations, and unless some action is taken "the whole genetic capacity of man will be much weakened."<sup>91</sup>

This bleak picture has prompted the suggestion of measures designed to cope with the deteriorating gene pool. Such measures rest on the belief that the present pattern of genetic selection is much less desirable than that which could be achieved by a deliberate and controlled effort. Two types of proposals are frequently suggested. The first is a program designed

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<sup>89</sup> Paul Ramsey, "Moral-Religious Implications of Genetic Control," in John D. Roslansky (ed.), Genetics and the Future of Man, (Appleton-Century-Crofts: New York, 1966), p. 111.

<sup>90</sup> Bentley Glass, "Human Heredity and Ethical Problems," Perspectives in Biology and Medicine, Vol. 15, Winter 1972, p. 243.

<sup>91</sup> Julian Huxley, quoted in Sol Tax and Charles Callender (eds.), Evolution After Darwin, Vol. III, (The University of Chicago Press: Chicago, 1960).

to alter genetic composition by encouraging desirable traits, i.e. "positive" eugenics.<sup>92</sup> The second is designed to alter genetic composition by reducing the incidence of undesirable traits, i.e. "negative" eugenics. The latter alternative might be accomplished by persuading those who have a high likelihood of transmitting a genetic defect not to reproduce, by sterilization, or by abortion of fetuses diagnosed as genetically abnormal.

The "deteriorating gene pool" argument is not without its critics. They contend that the predicted danger of a genetic apocalypse is erroneously calculated. They see no "imminent danger of being overwhelmed by the bad genes . . . we would seem to have no reason to fear that the normal population will soon be replaced by that of individuals with abnormal genetic factors."<sup>93</sup> One force working to reduce the genetic burden is current demographic trends. "In the short run, demographic trends (in and of themselves) are reducing the incidence of serious congenital anomalies."<sup>94</sup> Trends indicating smaller family size and a lowered average age of childbearing will work to ameliorate the quality of the human gene pool.

Another criticism concerns the nature and severity of genetic disease. "Many a 'bad' gene whose effects are overcome euphenically [i.e. by medical treatment] may be said to have lost its 'badness,' wholly or to a large degree so that its accumulation no longer represents a serious biological load even

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<sup>92</sup> See supra, n. 66, for a discussion of the implications of such programs.

<sup>93</sup> Reisman and Matheny, op. cit., supra, n. 15 at 204.

<sup>94</sup> Dudley Kirk, "Patterns of Survival and Reproduction in the United States," Proceedings of the National Academy of Sciences of the U.S.A., Vol. 59, March 1968, p. 669.



though it may represent a considerable economic load."<sup>95</sup> For example, if a disease such as diabetes can be controlled by artificially altering the environment, i.e. providing easy acquisition to insulin, then any real harm to individuals is negated. Thus, "environmental changes have made some hereditary defects irrelevant";<sup>96</sup> it seems reasonable to expect similar medical advances in the future.

There are those who also question an underlying assumption of proposals designed to "protect" the gene pool - that such protection is an obligation of this generation to future generations. Professor Martin P. Golding contends that "We are thus raising a question about our moral relations to the community of the remote future. I submit that this relationship is far from clear, certainly less clear than our moral obligations to communities of the present."<sup>97</sup> It seems highly unlikely that today's generation can accurately predict the needs and wants of succeeding generations.

One might go so far as to say that if we have an obligation to distant future generations it is an obligation not to plan for them. Not only do we not know their conditions of life, we also do not know whether they will maintain the same (or a similar) conception of the good life for man as we do.<sup>98</sup>

Even if it could be agreed that there are real and identifiable obligations

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<sup>95</sup> Philip Handler (ed.), Biology and the Future of Man, (Oxford University Press: New York, 1970), pp. 910-911.

<sup>96</sup> Kirk, op. cit., supra, n. 94.

<sup>97</sup> Martin P. Golding, "Ethical Issues in Biological Engineering," UCLA Law Review, Vol. 15, February 1968, p. 453.

<sup>98</sup> Martin P. Golding, "Obligations to Future Generations," The Monist, Vol. 56, January 1972, pp. 97-98.

to future generations, there is still the problem of deciding how to balance those obligations against the obligations to the present generation.

Any attempt to manipulate the existing gene pool might not only foreclose possible options of future generations, but might also adversely affect their biological adaptability. The genetic diversity of the human gene pool has long been recognized as necessary for ensuring adaptability to future environments, so essential to survival in the face of constant evolutionary change. "Genetic diversity is in one sense capital for investment in future adaptations. Since genetic variability represents evolutionary capability, it is a load we should be ready and willing to bear."<sup>99</sup> It would appear morally and biologically unwise, then, to tamper with the gene pool significantly without prior knowledge of the demands and needs of future environments.

Finally, many seriously doubt the efficacy of negative eugenic programs. Most deleterious genes are maintained in the population by normal heterozygous persons. It is estimated that "every individual is a carrier of three or more of such genes, and that virtually every human being carries at least one."<sup>100</sup> Since one cannot always be certain that such genes will manifest themselves clinically in present or future generations, "only by eliminating virtually everyone could our load of past mutations be eliminated, and this only temporarily, as new mutations are occurring all the time."<sup>101</sup>

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<sup>99</sup>Marc Lappe, "Moral Obligations and the Fallacies of 'Genetic Control,'" Theological Studies, Vol. 33, September 1972, p. 423.

<sup>100</sup>Arno G. Motulsky, George R. Fraser and Joseph Felsenstein, "Public Health and Long-Term Genetic Implications of Intrauterine Diagnosis and Selective Abortion," in Daniel Bergsma (ed.), Intrauterine Diagnosis (Birth Defects: Original Article Series, Vol. 7: The National Foundation - March of Dimes, April 1971), p. 26.

<sup>101</sup>Orlando J. Miller, "Discussion of Symposium Papers," Ibid., p. 34.

An alternative suggestion for improving the gene pool, without acting directly upon biological man, is to minimize or eliminate environmental hazards. It is becoming clear that "we are exposed to a wide range of chemical and physical agents which may damage the genetic material of our cells."<sup>102</sup> Thus, to the extent that man contaminates his environment and introduces factors that render it harmful, "his best interests are served by the adoption and enforcement of regulatory measures to prevent, minimize, or remove undesirable contamination."<sup>103</sup>

In view of the sometimes vehement stands taken by those on both sides of the argument, it would seem to be useful to begin to assess the status of the human gene pool. A six-year report of the American Eugenics Society speaks directly to this point:

In view of the relative stability of the gene pool, the problem is not generally viewed as one requiring dramatic or 'crash' solutions. But in the long run, changes in the distribution and frequencies of genes may be of greatest significance. At this stage the need is for better identification of the present and potential direction of changes rather than action to alter these trends in any major way.<sup>104</sup>

It is also not too soon to begin to evaluate some of the suggested approaches for improving man's capability to control genetic disease.

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<sup>102</sup>Bloom, op. cit., supra, n. 31 at 1. See also V.E. Headings, op. cit., supra, n. 50.

<sup>103</sup>National Commission on Community Health Services, Changing Environmental Hazards, Report of the Task Force on Environmental Health (Public Affairs Press, Washington, D.C., 1967), p. 20.

<sup>104</sup>Theodosius Dobzhansky, Dudley Kirk, Otis Duncan and Carl Bajema, The American Eugenics Society, Inc. Six-Year Report, 1965-1970 (Published by the Society: New York), p. 6.

## II. Prenatal Diagnosis and Selective Abortion

When prenatal diagnosis is combined with abortion, it becomes possible to alter directly the course of genetic disease. The introduction of new automated procedures will facilitate diagnosis as well as lessen its cost. It is likely, therefore, that as methods for intrauterine diagnosis improve, that pressures to use them in the management of the pregnant patient will increase.<sup>105</sup> Undoubtedly, this new technical capability will also add another dimension to the debate concerning abortion. It would be useful, therefore, to examine some of the criteria for developing policy for this alternative.

A. Risks/Benefits of Prenatal Diagnosis: One important consideration is the risks involved in using various prenatal diagnostic tests. All of the techniques described herein carry some degree of risk. While most of the evidence appears to indicate that the risk is minimal, much more data remains to be collected and evaluated. In the case of amniocentesis, for example, it has been suggested that "the current status of knowledge of the biology of amniotic fluid and its contents - including the fetus - is so rudimentary that this field must be regarded primarily as an area for research."<sup>106</sup> As noted earlier (supra, p. 7), very little information is available regarding the long-term risks of applying amniocentesis. Since the use of any diagnostic technique is justified only if the frequency of the disease or its severity

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<sup>105</sup> "As couples feel social pressure to limit population growth and to be content with only two children, most will very much want to ensure that the two they do have are healthy. I expect the demand for amniocentesis to develop strongly." C.O. Carter, "Practical Aspects of Early Diagnosis," in Maureen Harris (ed.), Early Diagnosis of Human Genetic Defects: Scientific and Ethical Considerations, Fogarty International Center Proceeding, No. 6, 1972. (Department of Health, Education and Welfare, Publication No. (NIH) 72-75), p. 20.

<sup>106</sup> Orlando J. Miller, op. cit., supra, n. 101 at 33.

are greater than the risks posed by the diagnostic procedure, a careful assessment of those risks and the reliability of the diagnosis should be made.

Because of the danger of applying these techniques to the general population before their costs - in terms of morbidity and mortality to mother and fetus - have been reliably assessed, it has been suggested that they be used only when a couple carries a moderate or high risk of giving birth to a child with a genetic defect.<sup>107</sup> Various criteria for making such determinations have been proposed.<sup>108</sup> For example, statistics indicate that increasing age at pregnancy is correlated with an increased incidence of chromosomal abnormalities.<sup>109</sup> Thus, prospective prenatal diagnosis might be warranted of mothers above a certain age.<sup>110</sup>

As familiarity with these techniques increases, the risks will surely diminish. Automation will make the required tests simple and inexpensive and more couples will undoubtedly request them. Thus, by emphasizing the criterion of risks/benefits, there might well come a time when prenatal diagnosis will be an integral part of monitoring all pregnancies. There are, however, other considerations.

B. Parent-Child Relationship: Since treatment or cure is not available for most genetic diseases, the only therapeutic alternative following the

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<sup>107</sup>Michael M. Kaback, quoted in Maureen Harris (ed.), op. cit., supra, n. 105 at 85.

<sup>108</sup>C.O. Carter, Ibid., pp. 18-19.

<sup>109</sup>Luks and Ruddle, op. cit., supra, n. 9 at 495-497.

<sup>110</sup>In the case of Down's syndrome, for example, a substantial proportion - as high as 33 per cent - of children are born to mothers above the age of 40 years. Motulsky, et. al., op. cit., supra, n. 100 at 30.

diagnosis of a defective child is an abortion. Important to consider here is the effect that the wide-scale application of prenatal diagnosis combined with abortion will have on the parent-child relationship. Ethicist John Fletcher contends that

the experience of parents in prenatal diagnosis and genetic counseling does not lessen the affection they bear for their children, already born or to be born, even though that relationship is permanently altered by the character of the experience of genetic counseling and amniocentesis.<sup>111</sup>  
(emphasis added)

In his efforts to identify and describe this "altered relationship,"<sup>112</sup> Fletcher suggests that a new stage of life is created, in which parents will be as intimate with their children before they are born as they are after they are born. One result of this was "that active roles as parents began earlier in the course of pregnancy . . . Assurance of the health of the child releases parental care, planning and symbolic activity usually reserved for birth."<sup>113</sup> Fletcher quotes a number of couples responding that "they loved them [their children] more because they had known them longer."<sup>114</sup> Ironically, Fletcher also found that this pre-natal intimacy "increases the sense of compulsion towards perfection that middle-class people have; they want their

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<sup>111</sup> John Fletcher, "The Brink: The Parent-Child Bond in the Genetic Revolution," Theological Studies, Vol. 33, September 1972, p. 428.

<sup>112</sup> Ibid., pp. 457-485. His sample consisted of 25 couples who had undergone amniocentesis and had given birth to a healthy child or had an abortion performed.

<sup>113</sup> Ibid., p. 477.

<sup>114</sup> Interview with Dr. John Fletcher, Director of Interfaith Metropolitan Theological Education, Inc., Washington, D.C., August 11, 1972.

babies to be healthy, beautiful and perfect."<sup>115</sup> He sees a danger in this, contending that "the drive towards perfection is one of the worst qualities that human beings have since it causes them to become very intolerant."<sup>116</sup>

Fletcher also asks if prenatal diagnosis,

because it inclines the parents to contemplate the abortion of the fetus before they are fully informed as to the results of the test, erode[s] that "basic trust" which is so fundamental as to lead Erik Erikson to assert that "the firm establishment of enduring trust over basic mistrust is the first task of the budding personality and therefore first of all a task for maternal care"?<sup>117</sup>

Fletcher contends that even if the diagnostic results are negative, the test and its results are going to change the lives of the parents. "They will never be the same parents they were before because this test is changing the way they learn the roles of parenthood."<sup>118</sup> People have been brought up to love their child, at least prior to its birth, without preconditions. But, says Fletcher, "when you start contemplating the tests with the added feature that abortion is an alternative, you have introduced an element of doubt into that relationship that has never been there before. So you are a different parent than you were taught to be."<sup>119</sup> Thus, genetic technology has altered parenthood in a way that had not been anticipated.

What will be the effect of this new dimension of parenthood on the "basic

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<sup>115</sup>Idem.

<sup>116</sup>Idem.

<sup>117</sup>Fletcher, op. cit., supra, n. 111 at 473.

<sup>118</sup>Interview with John Fletcher, supra, n. 114

<sup>119</sup>Idem.

trust" between child(ren) and parents? Fletcher states that "nobody can live with the thought that his parents would have killed him if he had been sick. There is no way to accept that, yet you are going to have a whole generation of parents who have had this opportunity."<sup>120</sup> In the context of its present usage, Fletcher believes that prenatal diagnosis "does not introduce a permanently insoluble moral conflict in the ethics of parental caring."<sup>121</sup> But what will be its effect if applied on a wide-scale and supported by socially and legally sanctioned abortion? Fletcher believes that

Nothing could weaken or dissolve the parent-child bond more effectively than children becoming afraid that the parents made such decisions for trivial reasons of personal convenience or because they were forced into it for external societal reasons.<sup>122</sup>

The parent-child relationship, then, constitutes another important variable to consider when developing genetic technology policy (see infra, pp. 63-64, for additional discussion of intrafamily relationships).

C. Economic Variables: Economic factors must also be considered. The economic impact can be evaluated on two levels: (1) the burden which falls on the individual family, and (2) the costs to society. The birth of a genetically defective child creates new problems of resource allocation for a family. The cost of caring for such a child can make deep inroads into a family's financial

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<sup>120</sup> Idem. Fletcher also inquires into the feelings of living children, e.g. will they worry about their own security?, where amniocentesis was used on a fetal sibling. Op. cit., supra, n. 111 at 478.

<sup>121</sup> Fletcher, op. cit., supra, n. 111 at 479.

<sup>122</sup> Ibid., p. 480.



resources (witness the case of cystic fibrosis, supra, p. 23) and private health insurance has been unable to absorb this impact. This drain on resources might also have disruptive consequences for the family unit in more subtle ways.<sup>123</sup>

The costs to society are both direct and indirect. Society not only assists in providing care for defective individuals, but also assumes the losses resulting from their inability to become economically and socially productive members of society. Institutionalization and care for persons with genetic disorders, many of which are chronic in nature, can be very expensive. For example, the cost to society of caring for those suffering from Down's syndrome, which has an estimated frequency of one in 600 births, is approximately \$1.7 billion annually.<sup>124</sup> The economic impact of genetic disease, then, must be weighed along with other factors.

D. Abortion and the Gene Pool: While a program of selective abortion might help to relieve the emotional and financial strain experienced by individuals and their families, how effective would such a program be in reducing the total frequency of deleterious genes? Arno Motulsky and his colleagues have found that the use of selective abortion to reduce the cases of autosomal recessive diseases "will be relatively small (between 12.5% and 34%) if the procedure is only initiated following birth of an affected child."<sup>125</sup> In fact, selection against recessive genes under any conditions

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<sup>123</sup> See McCollum, op. cit., supra, n. 73 at 1335-40, for a discussion of some of the educational, social and psychological needs of other family members that might be compromised by the strain on family resources.

<sup>124</sup> Glass, op. cit., supra, n. 90 at 241.

<sup>125</sup> Motulsky, et. al., op. cit., supra, n. 100 at 30.

will be ineffective unless there is also selection against heterozygotes. For example, "a 50% reduction in reproduction of heterozygotes would reduce the incidence of the recessive homozygote under random mating to one fourth its former value in one generation."<sup>126</sup> For maximum effectiveness in eliminating autosomal recessive diseases, premarital carrier detection would be required to detect those matings at risk.<sup>127</sup>

Sex-linked diseases can be prevented by prenatal diagnosis of heterozygote mothers and selective abortion of all male fetuses. The impact of such a program, however, would be somewhat softened since many harmful sex-linked diseases are a result of fresh mutations. Thus, "even with prospective diagnosis, the maximum case reduction would not exceed two-thirds of existing affected males for diseases with zero fertility."<sup>128</sup> A potential dysgenic effect of such a program is that abortion of all male fetuses of heterozygote mothers would result in an increase (as much as 50 per cent with each generation in the case of hemophilia)<sup>129</sup> in female carriers in future generations, thus requiring more abortions.

The possibility that selection against autosomal recessive diseases would lead to an increase in their gene frequency could be the result of "reproductive compensation," since couples would be inclined to replace the affected child lost by abortion. A proportion of these compensating children

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<sup>126</sup>James F. Crow, "Rates of Genetic Change Under Selection," Proceedings of the National Academy of Sciences of the U.S.A., Vol. 59 March 1968, p.660.

<sup>127</sup>Motulsky, et. al., op. cit., supra, n. 100 at 30.

<sup>128</sup>Ibid., p. 31.

<sup>129</sup>Friedmann, op. cit., supra, n. 11 at 40.

will be abnormal gene carriers, thus increasing the frequency of abnormal genes over that which would have resulted had no such program been implemented. There is evidence, however, that "despite compensation, the total effects on gene frequency are minimal and are not a cause for concern."<sup>130</sup>

A program of selective abortion aimed at reducing the frequency of harmful genes raises a number of sensitive issues. For example, if a distinction between affected fetuses and clinically normal carriers cannot be made, as in the case of hemophilia, half of the male fetuses aborted would be normal. The moral implications of such a procedure must be weighed along with other considerations. A concomitant problem resulting from the inability to distinguish between affected and normal male fetuses in utero is that the result would be a 75 per cent probability of abortion with each pregnancy. This would mean a "24% risk that five consecutive pregnancies would be aborted."<sup>131</sup> In this instance, then, the deleterious effect on the couple involved might be greater than if no such program were introduced.

As noted earlier, abortion of all male fetuses where sex-linked diseases are indicated would result in an increase in female carriers, thus increasing the frequency of the harmful gene and the need for abortion. Is there also justification for aborting female carriers? The abortion of such fetuses is morally questionable since they exhibit no clinical manifestation of the disease. From a population and public health point of view, a recent study found little evidence to support such a program.<sup>132</sup> Perhaps such carriers

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<sup>130</sup> Motulsky, et. al., op. cit., supra, n. 100 at 31.

<sup>131</sup> Michael M. Kaback, "Discussion of Symposium Papers," Ibid., p. 35.

<sup>132</sup> Motulsky, et. al., op. cit., supra, n. 103 at 27.

could be counseled not to reproduce; in this event, the moral and political overtones of such a policy need to be carefully assessed. Finally, there are those who believe that the development of new medical techniques for treating such diseases will make the abortion of such fetuses unnecessary.<sup>133</sup>

E. Attitudes and Policies on Abortion: Another factor which will influence the introduction and development of this approach involves existing attitudes and policies concerning abortion. In a study of 25 couples, Fletcher found that while "abortion is the major moral problem of parents in genetic counseling, [they] are inclined to favor abortion in case of a positive diagnosis, and they have reached this position prior to counseling."<sup>134</sup> On a much broader scale, a recent national survey found that majority support for legal abortion has increased sharply. The survey revealed that 64 per cent of all Americans support full liberalization of abortion laws, believing that "abortion should be a matter for decision solely between a woman and her physician."<sup>135</sup> Recent statements by both public and private groups also reflect a more liberal attitude toward abortion. For example, the Commission on Population Growth has recommended that "present state laws restricting abortion be liberalized along the lines of the New York statute [which, prior

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<sup>133</sup> Fritz Fuchs, quoted in Maureen Harris (ed.), op. cit., supra, n. 108 at 124-125. For example, ten years ago the chances were remote that a baby with Down's syndrome would live beyond its 15th birthday. Since that time, however, the development of new antibiotics has given such children a projected life expectancy of 50 years or more. See Joseph D. Whitaker, "Science Lends Hand to Mongoloid Baby," Washington Post, December 18, 1972, p. A1

<sup>134</sup> John Fletcher, "Moral Problems in Genetic Counseling," Pastoral Psychology, April 1972, p. 60.

<sup>135</sup> George Gallup, "Abortion Seen Up to Woman, Doctor," Washington Post, August 25, 1972, p. A2.

to the recent Supreme Court ruling, was the most liberal of the state abortion laws]." <sup>136</sup> And the World Council of Churches has called for its members

to be prepared to endorse the personal right of parents to choose an induced abortion to prevent the birth of a gravely defective child. Wherever the laws of the state make this illegal, the churches should press for a modification of the law to permit such options to take place.<sup>137</sup>

The possibility of using therapeutic abortions for genetic purposes raises concern among many who fear that in a social climate in which unwanted pregnancy is sufficient indication for abortion, there will be a tendency for couples to seek abortions for arbitrary and casual reasons.

With increasing acceptance of abortion and limitations on family size, it is probable that some families will seek termination of pregnancies that involve less severely affected fetuses, or those with disorders that are treatable to some extent . . . . It is also likely that abortion may be chosen for disorders of uncertain severity. It can in fact be anticipated that families will not want to risk any departure from the normal karyotype in their offspring.<sup>138</sup>

Perhaps it is appropriate to recall Fletcher's fears regarding the possible growth in intolerance on the part of future parents (supra, pp. 36-37). Those who fear the emergence of an "abortion mentality," characterized by an increasing intolerance for "weakness" or differentiation from a given "norm,"

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<sup>136</sup> The Report of the Commission on Population Growth and the American Future, Population and the American Future, (U.S. Government Printing Office: Washington, D.C., March 1972), p. 142.

<sup>137</sup> Working Committee on Church and Society, op. cit., supra, n. 4 at 6.

<sup>138</sup> Aubrey Milunsky, John W. Littlefield, Julian N. Kanfer, Edwin H. Kolodny, Vivian E. Shih and Leonard Atkins, "Prenatal Genetic Diagnosis (Third of Three Parts)," The New England Journal of Medicine, Vol. 283, December 31, 1970, p. 1502. The authors cite the XYY chromosome anomaly as a disorder of "uncertain severity." The varied scientific opinion regarding this genetic defect and its consequent policy implications will be discussed later.

point to an almost casual acceptance of abortion. This attitude, they maintain, is reflected in statistics from those states which had liberal abortion laws and in the growing acceptance of "early-stage abortion," which allows a woman with a suspected and unwanted pregnancy simply to have her monthly menstrual period extracted.<sup>139</sup> This concern over the effect of a program combining prenatal diagnosis with selective abortion may be expressed as important questions for policy: Do acceptable standards for deciding when to abort need to be established? Whose responsibility is it to develop and apply those standards? What will be the effect of such a program on attitudes toward already existing "genetically defective" children?

F. Policy Alternatives: Debate has already begun regarding the types of policy adjustments that might be made. For example, should a woman be required to agree to an abortion prior to prenatal diagnosis? Some contend that "For parents unwilling to take that step, diagnosis of a disease in a fetus would serve no useful purpose and would only create anxiety and grief for the parents."<sup>140</sup> Thus, they firmly believe that "the decision to interrupt the pregnancy, if the suspected disorder is verified in the fetus, should be made before the amniocentesis."<sup>141</sup> Should there be special provisions, however, for those patients or physicians whose religious convictions preclude an

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<sup>139</sup> For details of the procedure and some of its problems, see Time, September 11, 1972, p. 47; also the Washington Post, January 26, 1973, p. C5.

<sup>140</sup> Arno G. Motulsky, "Genetic Therapy: A Clinical Geneticist's Response," in Michael Hamilton (ed.), op. cit., supra, n. 86 at 131.

<sup>141</sup> Fritz Fuchs, "Amniocentesis and Abortion: Methods and Risks," op. cit., supra, n. 100 at 19.

abortion under any circumstances?<sup>142</sup> There are others who find unacceptable such restrictions on individual decision-making. The use of prenatal diagnosis does not mean that "the geneticist may abrogate the couple's decision by assuming that if the fetus is normal she will carry it, or if abnormal, she will abort. The genetic component is one of many, and the client must be helped to put it in perspective for a positive choice."<sup>143</sup>

With respect to the difficult problem of deciding how "abnormal" a fetus must be to justify abortion, one geneticist has suggested that society must take advantage of "all morally acceptable developments that promise to minimize the number of unfortunate individuals incapable of full participation in this complex society."<sup>144</sup> While this position might attract sympathy, it would probably draw an equal amount of skeptical criticism. How is one to determine if a fetus will be "incapable of full participation"? This is a very real problem, amply illustrated by the case of Down's syndrome

Some Down's children have rather gross retardation, major heart anomalies, and many fail to survive infancy; on the other hand, some have a rather mild retardation . . . no major heart defects, and have lived to at least middle age. An individual carrying a gene or genes which cause

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<sup>142</sup>Of relevance here is a resolution (S.J. Res. 64) recently introduced by Senator Frank Church, which would make it national policy, in the administration of all Federal programs, to protect physicians and health care personnel in their exercise of religious or philosophical beliefs which proscribe the performance of abortions or sterilization procedures. Congressional Record, February 15, 1973, pp. S2567-68.

<sup>143</sup>E. James Lieberman, "Psychological Aspects of Selective Abortion," Ibid., p. 20.

<sup>144</sup>James V. Neel, "Lessons from a 'Primitive People,'" Science, Vol. 170 November 20, 1970, pp. 820-21.

retardation may be more or less retarded depending on other genetic factors and the external environment which is at work.<sup>145</sup>

Furthermore, many Down's children have been found not to suffer and to have good emotional adjustment.<sup>146</sup> Thus, it would be difficult to determine an absolute measure of biological fitness, since such fitness is to some extent dependent on a particular environment.

A policy question which pervades all others concerns the basis upon which society will allocate decisions to either personal conscience or public choice. At what point is society's intervention into individual decision-making justified? As prenatal diagnosis becomes more widespread, the tensions resulting from its application will become more acute. There is a need to relieve those tensions, balancing individual and societal needs with the proper respect for human life.

### III. Screening for Genetic Disease

If prenatal diagnosis and selective abortion were combined with screening programs designed to detect heterozygous carriers, it might be possible to realize significant reductions in the incidence of some recessive diseases. If at-risk parents were identified prior to reproduction, they could eliminate the risk by remaining childless, by adopting their family, or, when available, by artificial insemination or prenatal diagnosis. Another advantage of such programs is that the detection of the homozygous child after birth might be followed by immediate treatment, thus reducing and perhaps eliminating the deleterious effects of the disease.

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<sup>145</sup>Robert C. Baumiller, "XYY Chromosome Genetics," Journal of Forensic Sciences, Vol. 14, October 1969, p. 417.

<sup>146</sup>Karen Lebacqz, Letter to the Editor, The Hastings Center Report, Vol. 2, February 1972, pp. 12-13.



A. Cost/Benefits of Screening: Cost/benefit analysis has shown that screening programs would result in large savings for both the family and society. For example,

The cost for successful medical treatment of phenylketonuria is estimated to be no more than one tenth the cost of care for a retarded patient in an institution. Early diagnosis and treatment thus saves the community about \$9,000 annually per patient. Moreover, the patient who escapes the immediate consequences of this mutant allele will eventually earn income and pay taxes, representing a further benefit to the community.<sup>147</sup>

In the case of detection prior to birth, a recent study demonstrated that in the case of cystic fibrosis, a substantially favorable economic ratio would result.<sup>148</sup> As new screening techniques become available an important part of planning large-scale screening programs should be the assessment of the costs involved in treating the genetic diseases. Present evidence seems to indicate a substantial economic saving.

B. Screening and the Gene Pool: Another important criteria for assessing the value of screening is its potential for reducing the frequency of deleterious genes. The underlying assumption of such a program is that heterozygous couples will not mate, or in instances where they do, they will not have their own children. It has been suggested that this latter alternative is best realized through a program of voluntary sterilization. It should be useful, therefore, to review the potential impact of these two approaches on the gene pool.

If the fertility of heterozygotes and normal individuals were identical,

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<sup>147</sup> Charles R. Scriver, "Screening for Inherited Traits: Perspectives," in Maureen Harris (ed.), op. cit., supra, n. 105 at 95-96.

<sup>148</sup> Motulsky, et. al., op. cit., supra, n. 100 at 30.

the frequency of the abnormal gene would remain constant. However, many genes which result in autosomal recessive diseases owe their high frequency to the heterozygote's advantage in fertility or mortality. If heterozygotes were to cease mating with one another and this advantage persisted, there would be an increase in the genes since heterozygotes would have a greater average number of children. Consequently, the

gene loss previously incurred by infertility or early death in homozygotes would cease. In the case of cystic fibrosis persistence of the assumed heterozygote advantage for about 100 generations would increase the frequency of carriers in white populations from 5% (its present level) to 50%.<sup>149</sup>

If there were little or no difference in fertility between heterozygotes and normal persons, as might be the case if family size became more standardized, a system in which heterozygotes avoided marriage would prevent a decrease in abnormal gene frequency. There might even be a slow increase due to fresh mutations, though several thousand generations would pass before the frequency of carriers would be doubled.<sup>150</sup>

Sterilization of heterozygous carriers appears unlikely to have any substantial impact on reducing deleterious genes. If the program were compulsory, it would require 1,500 years to reduce the frequency of a particular recessive gene by half.<sup>151</sup> If sterilization is undertaken on a voluntary basis, the rate of decrease would be much less. Thus, the elimination of a recessive defect by sterilization is a very slow process and probably of no immediate

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<sup>149</sup> Ibid., p. 28.

<sup>150</sup> Ibid.

<sup>151</sup> Ching Chun Li, Population Genetics, (The University of Chicago Press: Chicago, 1955), p. 253.

value in eugenic programs.

C. Voluntary versus Compulsory Screening: Genetic screening raises other essential policy issues. One crucial and heavily debated issue concerns the nature of such programs: Should participation in screening programs be voluntary or be made compulsory? The controversy over this question has turned into a full-fledged debate. At least ten states<sup>152</sup> and the District of Columbia have enacted screening programs which will either require, or at the discretion of a doctor or health officer may require, black persons to undergo tests for sickle-cell anemia. There is little disagreement about the desirability of such tests if they are voluntary, but when the tests are made mandatory, the debate becomes vigorous.

The City Councilman who introduced the compulsory sickle-cell anemia legislation in the District of Columbia defends his position, contending that "this is a trait and a disease that has been ignored. There is no cure, but a family knowing the facts would know what counseling or steps to take . . . I don't think we can get at the problem on a voluntary basis. There is too much apathy."<sup>153</sup> On the other side of the debate, there are many persons who find mandatory programs both unnecessary and counter-productive. A recent genetics task force of the Institute of Society, Ethics, and the Life Sciences strongly urged that

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<sup>152</sup>The ten states are: Georgia, Illinois, Indiana, Iowa, Kentucky, Louisiana, Maryland, Massachusetts, New York and Virginia. At least four of these states - Maryland, Massachusetts, New York, Virginia - and the District of Columbia will consider legislation in 1973 to repeal the compulsory features of their programs.

<sup>153</sup>Henry S. Robinson, quoted in Victor Cohn, "Disease Publicity Raises Problems." Washington Post, November 12, 1972, p. A12.

genetic screening programs should be conducted on a voluntary basis . . . . There is currently no public-health justification for mandatory screening for the prevention of genetic disease. The conditions being tested for in screening programs are neither "contagious" nor, for the most part, susceptible to treatment at present.<sup>154</sup>

It is also feared that state enforced screening programs will be the beginning of greater government intervention into what many consider to be an area for private decision-making.

When you start talking about compulsory testing, you also start talking about compulsory genetic counseling. When you start talking about compulsory genetic counseling, you start talking about putting the state behind it. Then you get into all sorts of implications . . . . I'm for voluntary sickle trait testing, but I believe compulsory genetic testing sets a bad precedent in our kind of society.<sup>155</sup>

The possible implications of compulsory counseling might include state-supported marriage and sterilization laws for genetic purposes, the precedents for which already exist. In fact, 25 states still retain eugenic sterilization statutes, 22 of which are compulsory.<sup>156</sup> There are also state laws prohibiting

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<sup>154</sup> A report from the Research Group on Ethical, Social and Legal Issues in Genetic Counseling and Genetic Engineering of the Institute of Society, Ethics and the Life Sciences, "Ethical and Social Issues in Screening for Genetic Disease," The New England Journal of Medicine, Vol. 286, May 25, 1972, pp. 1130-31. In the case of sickle-cell anemia there is no acceptable and effective treatment at this time. See R.B. Scott, "Urea Therapy in Sickle-Cell Anemia," The New England Journal of Medicine, 285: 1025-26, October 28, 1971.

<sup>155</sup> Paul McCurdy, Georgetown University, quoted in Victor Cohn, op. cit., supra, n. 153.

<sup>156</sup> William R. Matoush, "Eugenic Sterilization - A Scientific Analysis," Denver Law Journal, Vol. 46, 1969, p. 633. In recent years, however, these laws have not been enforced.

consanguineous marriages, most prohibiting marriage between first cousins or persons more closely related.<sup>157</sup> Might not the same legal rationale which led to these laws also be used to justify their application to carriers of deleterious genes?<sup>158</sup> While a definitive answer is not possible at this time, there is some speculation that all such laws might be declared unconstitutional. In Griswold v. Connecticut,<sup>159</sup> the Supreme Court held that the state has no power to interfere with the use of contraceptives by married couples, such use being considered one of the rights reserved to the people under the Ninth Amendment. If one interprets the case broadly, it may be read "to affirm that the decision by a husband and wife to have children, or not to have children, or how many children to have, is one in which the state may not interfere, whether the purpose be to limit the population or to improve it eugenically."<sup>160</sup> Underlying this reasoning are certain assumptions regarding the "rights" of couples to reproduce.

D. Procreation and Genetic Disease: If procreation is viewed as a couple's "right," then it should be useful for planning genetic counseling activities and services to have some feeling for the kinds of reproductive decisions that couples will make and the reasons for their decisions. Ramsey

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<sup>157</sup> Michael Farrow and Richard Juberg, "Genetics and Laws 'Prohibiting' Marriage in the United States," Journal of the American Medical Association, 209: 535-538, July 28, 1969.

<sup>158</sup> In this regard it is interesting to note that Virginia's law also requires sickle-cell screening for marriage license applicants. Virginia Code, section 32-112.14

<sup>159</sup> 381 U.S. 479 (1965).

<sup>160</sup> Frank P. Grad, "Legislative Responses to the New Biology: Limits and Possibilities," UCLA Law Review, Vol. 15, February 1968, p. 488.

finds it "shocking to learn . . . how many parents will accept grave risk of having defective children rather than remain childless."<sup>161</sup> Unfortunately, there are only a few empirical studies concerning the tendency toward risk-taking among couples and the results are somewhat mixed. In his study of 25 couples, Fletcher found that "Given the choice of accepting a genetically defective child or resorting to abortion, . . . they would choose the latter."<sup>162</sup> Another study involving 455 couples found that "on the whole, they took responsible decisions on the basis of the information. Where the recurrence risk was high - that is, equal to or greater than 1 in 10 - two-thirds (109 out of 170) were deterred from planning further children."<sup>163</sup> In his study at Yale, Hsia reports that only 25 per cent of the couples in a high risk group were deterred by counseling.<sup>164</sup> And in their study of 76 families, Leonard and his colleagues report that 34 (45 per cent) stated that "They regarded the disease as a reason for curtailing reproduction."<sup>165</sup> Finally,

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<sup>161</sup>Ramsey, *op. cit.*, *supra*, n. 89 at 166.

<sup>162</sup>Fletcher, *op. cit.*, *supra*, n. 134 at 53-54.

<sup>163</sup>C.O. Carter, K.A. Evans, J.A. Fraser-Roberts and A.R. Buck, "Genetic Clinic: A Follow-Up," *Lancet*, Vol. 1, February 6, 1971, p. 281. Both this and the Fletcher studies may be somewhat biased due to the character of the sample populations. The majority of the Fletcher sample was middle-class and has a graduate degree, while the Carter, *et. al.*, sample over-represented the upper social classes and was probably above average in education.

<sup>164</sup>Y.E. Hsia, "Choosing My Children's Genes," Paper presented at the American Association for the Advancement of Science annual meeting, Washington, D.C., December 29, 1972.

<sup>165</sup>Claire O. Leonard, Gary A. Chase and Barton Childs, "Genetic Counseling: A Consumer's View," *The New England Journal of Medicine*, Vol. 287, August 31, 1972, p. 435. This study focused on parents of children with three types of genetic diseases: cystic fibrosis, phenylketonuria, and Down's syndrome.

the results of a follow-up study<sup>166</sup> in England of 53 women referred for genetic counseling in families with Duchenne muscular dystrophy (a sex-linked disease for which there is no treatment) can be summarized as follows: Of the 41 women at high risk (defined as greater than 1 in 10), 36 decided to have no further children and two decided upon selective abortion. Only two disregarded the risks and intentionally became pregnant. Of the five women at medium risk (1 in 10 to 1 in 20), only one planned to have further children. The others considered the risks too great. Of the seven women at low risk (less than 1 in 20) only one was not reassured by the low risks and has avoided pregnancy.<sup>167</sup>

It should be pointed out that in most cases these studies refer to the impact of genetic counseling on couples' reproductive intentions, not their observed reproductive behavior. Generally speaking, these data suggest that counseling can have a significant impact on reproductive attitudes. The findings regarding actual reproductive behavior are not so encouraging. In the largest of the aforementioned studies, Carter and his colleagues report that of those high-risk couples who stated that they were attitudinally deterred from having further children, 24 per cent (26 out of 109) had at least one additional pregnancy. Among the low-risk couples, who also claimed to be attitudinally deterred, 15 per cent (9 out of 60) had at least one

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<sup>166</sup> A.E.H. Emery, M.S. Watt and E.R. Clack, "The Effects of Genetic Counselling in Duchenne Muscular Dystrophy," *Clinical Genetics*, 3:147-150, 1972. The investigators report that all social classes were represented and that their distribution was similar to that of the general population.

<sup>167</sup> Ibid., pp. 148 and 149.

additional pregnancy.<sup>168</sup> At least in this particular study, there appears to be a considerable degree of difference between the impact of genetic counseling on reproductive attitudes and its impact on actual reproductive behavior. There is an obvious need for more systematic investigation into the question of risk-taking as well as for determining those factors which influence such reproductive decisions. James R. Sorenson has identified some of these factors: "(1) the size of the risk, (2) the severity of the potential abnormality, (3) the social and private attitudes of the parents toward abnormality, (4) the economic capacity of the family to endure the burden of a genetic disease, (5) the genetic health of existing children, and (6) the type of counseling parents receive."<sup>169</sup> But as he points out, there is little data concerning the specific role that each of these factors (and perhaps other factors as well) plays in parental decision-making.

There is no consensus, however, that there is, or should be, a "right to procreation." Ramsey believes that

If the fact situation disclosed by the science of genetics can prove that a given person cannot be the progenitor of healthy individuals (or at least not unduly defective individuals) in the next generations, then such a person's "right to have children" becomes his duty not to do so, or to have fewer children than he might want (since he never had any right to have children simply for his own sake).<sup>170</sup>

Thus, Ramsey calls for the development and adoption of an "ethics of genetic

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<sup>168</sup> Carter, et. al., op. cit., supra, n. 163 at 283.

<sup>169</sup> James R. Sorenson, "Social Aspects of Applied Human Genetics," Social Science Frontiers, 1971, No. 3, Copyright (c) 1971 by Russell Sage Foundation, New York, p. 13.

<sup>170</sup> Ramsey, op. cit., supra, n. 87 at 35.



duty," whereby couples act responsibly and morally in order to prevent the birth of a defective child. Joseph Fletcher suggests that a more appropriate guideline for developing policy is "needs." He explains that

Needs are the moral stabilizers, not rights . . . . If human rights conflict with human needs, let needs prevail. If medical care can use genetic controls preventively to protect people from disease or deformity, or to ameliorate such things, then let so-called "rights" to be born step aside.

Rights are nothing but a formal recognition by society of certain human needs, and as needs change with changing conditions so rights should change too. The right to conceive and bear children has to stop short of knowingly making crippled children - and genetics gives us that knowledge . . .<sup>171</sup>

To what extent the state should be the agent for balancing the genetic "rights" and "needs" of its people is a question that society may soon have to face. There may be a fine line between a particular genetic defect being reason for a couple to refrain from procreation and its being reason for compulsory restrictions on the part of the state.

E. Target Populations: The nature of the program also raises questions concerning the populations toward which such programs should be targeted. The programs aimed at sickle-cell anemia clearly demonstrate the problems involved. These programs, and their enacting legislation, represent the nation's first genetic effort directed at a particular race.<sup>172</sup> While many other groups experience a high incidence of genetic disease, e.g. the Ashknazi

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<sup>171</sup> Joseph Fletcher, "Ethical Aspects of Genetic Controls: Designed Genetic Changes in Man," The New England Journal of Medicine, Vol. 285, September 30, 1972, p. 782.

<sup>172</sup> Sickle-cell anemia, with a frequency of 1 in 400, is the most common genetic disease in the black population. The incidence among the white population is much smaller. See Victor Cohn, "Disease's Effects Often Exaggerated," Washington Post, November 13, 1972, p. A8.

Jews and Tay-Sachs disease, and those of Mediterranean ancestry and Cooley's Anemia, none has been singled out for compulsory testing. The problem with isolating a specific ethnic group is that it might be interpreted as a racist gesture. Such has been the case with sickle-cell anemia, with some comparing it to the "racist eugenics legislation that led to the final solution in Nazi Germany,"<sup>173</sup> and others viewing it, when combined with some forms of genetic counseling, as "white genocide."<sup>174</sup> Whether or not these criticisms are valid, they are one reason for the growing opposition among blacks to sickle-cell programs. And yet, without their involvement and cooperation it is unlikely that such programs can accomplish their aims.

There is also criticism of laws such as those in Virginia, which require the screening of persons in correctional institutions and state mental hospitals. Some question the intent of such laws, maintaining that there is

no valid reason why prisoners and mental patients should be tested . . . the potential for mischief is great . . . scientific knowledge has in the past been perverted to fulfill social ends, and there is, unfortunately, nothing . . . which would lead one to believe there is no basis for alarm.<sup>175</sup>

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<sup>173</sup> James E. Bowman, Director of Laboratories, University of Chicago, quoted in Victor Cohn, op. cit., supra, n. 153.

<sup>174</sup> Victor Cohn, "Sickle-Cell Project Outlined," Washington Post, July 21, 1972, p. A15. A recent study reports a direct "relationship between fears of racial genocide and the use of family planning methods." The investigators note the depth and source of this fear, writing that Negroes are "responding to a long history of every possible type of oppression which has been perpetrated against blacks. The resistance to family planning and to family planning agencies run by whites is merely a symptom of the deep sense of historical and life-long estrangement." William Darity and Castellano Turner, "Family Planning, Race Consciousness and the Fear of Race Genocide," The American Journal of Public Health, Vol. 62, November 1972, pp. 1458-59.

<sup>175</sup> Bowman, quoted in Victor Cohn, op. cit., supra, n. 153.

Another concern for screening legislation is the age at which persons should be screened. For example, the District of Columbia requires that "Each child admitted to a public school, either kindergarten or the first grade as the case may be, shall have been tested for sickle-cell anemia."<sup>176</sup> There are many, however, who believe that testing at such an early age is of dubious value and probably undesirable. They argue that these children "are too young to fully understand the implications of being a trait carrier, could suffer from the stigma, and may forget all about it by the time they are likely to be considering marriage and child-bearing."<sup>177</sup> Much more data needs to be collected regarding the "best time" at which to initiate such testing.

F. Program Design and Management: Another broad policy concern is the implementation and administration of screening programs. It is essential that screening programs be designed for the purpose of attaining one or more predetermined goals. Establishing clearly defined goals will help to avoid circumstances which might be costly in both scientific and human terms. A recent report suggests that the most important goals of a screening program are those that

either contribute to improving the health of persons who suffer from genetic disorders, or allow carriers for a given variant gene to make informed choices regarding reproduction, or move toward alleviating the anxieties of families and communities faced with the prospect of serious genetic disease.<sup>178</sup>

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<sup>176</sup> Regulation No. 72-9, section 2 (May 5, 1972).

<sup>177</sup> B.J. Culliton, "Sickle-Cell Anemia: National Program Raises Problems As Well As Hopes," Science, Vol. 178, October 20, 1972, p. 284. Also, E. Beutler, D.R. Boggs, P. Heller, A. Maurer, A.G. Motulsky, and T.W. Sheehy, "Hazards of Indiscriminate Screening for Sickling," The New England Journal of Medicine, 285: 1485-86, December 23, 1971.

<sup>178</sup> Institute of Society, Ethics and the Life Sciences, op. cit., supra, n. 154 at 1129.

Another reason for establishing goals is to assist in program evaluation. If screening programs are to compete successfully with other programs for resource allocation, it will be necessary to provide "proof" of their effectiveness in order to justify public support. Identifiable goals are clearly needed for such evaluation. "Evaluation cannot exist in a vacuum. One must always ask evaluation 'of what.' Every action, every program has some value for some purpose."<sup>179</sup> Thus, an important task for program planners and administrators will be to develop appropriate measures and techniques for evaluating their efforts.<sup>180</sup>

The design and operation of screening programs raises other important considerations for policy-makers. One general observation concerns the relationship between public programs such as genetic screening and the communities to be served. Citizen pressure is becoming more influential in determining what services the community will receive. Suchman writes that

Once sufficient evidence has accumulated to indicate the potential benefits of a program, the public is likely to demand the program without waiting for conclusive proof. The greater the need, the stronger the pressure to put the program into operation as soon as it begins to look successful.

Thus, "popular causes" spring up which bring pressure upon the program administrator to satisfy public demand regardless of professional judgment or evaluation findings.<sup>181</sup>

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<sup>179</sup> Edward A. Suchman, Evaluative Research: Principles and Practice in Public Service and Social Action Programs, (Russell Sage Foundation: New York, 1967), pp. 37-38.

<sup>180</sup> For a relevant discussion of the needs and problems of program evaluation, see Suchman, Ibid., especially chapters 6-8; Aaron Wildavsky, "The Self-Evaluating Organization," Public Administration Review, 32:509-520, September/October 1972; and Thomas A. Morehouse, "Program Evaluation: Social Research Versus Public Policy," Ibid., 32:868-874, November/December 1972.

<sup>181</sup> Suchman, op. cit., supra, n. 179 at 153 and 152.

The difficulties involved are illustrated by the history of PKU legislation, in which "a small group of determined and highly motivated parents of mentally retarded children, together with a few equally dedicated physicians, needed less than three years to persuade forty-one states to pass laws requiring the testing of newborn children for phenylketonuria . . . ." <sup>182</sup> This effort has been characterized as "a simplified and incomplete understanding of the objective situation, a singleminded campaign which trumpeted success and ignored failures, and most of all a failure to consider the harm that might be done by seeking to do good." <sup>183</sup> The result has been a "poor piece of legislation, one with noble aims, but based upon unwarranted medical assumptions." <sup>184</sup> If the circumstances surrounding the evolution of PKU legislation are studied carefully, it may be possible to avoid similar pitfalls in planning future screening programs. <sup>185</sup>

G. Screening Tests: Extra care must be taken to develop testing procedures that will be accurate and subject to a minimum latitude of interpretation. A problem which might develop as a result of unreliable testing methods is that a "high proportion of false negatives or false positives not only will cast suspicion and discredit on the method, . . . but may result in professional

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<sup>182</sup> Bessman and Swazey, op. cit., supra, n. 47 at 49.

<sup>183</sup> Ibid., p. 50.

<sup>184</sup> American Academy of Pediatrics, "New Child Health Legislative Bills Proposed - Academy Subcommittee Issues Guidelines," quoted in Bessman and Swazey, Ibid., P. 72.

<sup>185</sup> History may already be repeating itself. There are cries that a new kind of "sickle-cell crisis," one due to hastily drawn and poorly-planned sickle-cell legislation, is occurring. See Cohn, op. cit., supra, n. 153 at A1.

malpractice charges . . . ." <sup>186</sup> The case of PKU illustrates this problem as well as more serious consequences.

Laboratory tests do not detect PKU but rather high blood phenylalanine levels, which can have causes other than PKU. Furthermore, the tests suitable for the mass screening required by law are subject to misinterpretation and error.

The tests are not accurate; they miss a number of cases of PKU and yield false positive reactions in an even greater number. Given a positive test, the physician will very probably put the child on a low phenylalanine diet . . . But a child who does not have PKU is actively endangered by the diet and can suffer physical deterioration at the least; a number of children have died from being treated for PKU, and it is likely that they did not have the disease.<sup>187</sup>

While it may be claimed that the physician is still able to decide the appropriate course of treatment for an infant, the fact that such legislation is a matter of public policy exerts a "powerful stimulus to prescribe in accordance with the cultural mores"<sup>188</sup> and, in the case of PKU, has resulted in the use of an unproven treatment. Reliable testing procedures, therefore, are necessary both to assure proper treatment and to gain the confidence and cooperation of the community.

H. Screening Services and Delivery: There is also the question of what services should be included in the designing of screening programs. At a minimum, such services should include follow-up diagnosis, treatment and

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<sup>186</sup> Irving Ladimer, "Legal Consideration in Screening, Treatment, Counseling and Research in Sickle Cell Disease," Paper presented at a Symposium on Sickle Cell Disease, New York, November 19, 1971, pp. 7-8.

<sup>187</sup> Bessman and Swazey, op. cit., supra, n. 47 at 50-51.

<sup>188</sup> Joseph D. Cooper, "The Role of Government Legislation in Management of Problems in Medicine," in Anderson and Swaiman (eds.), op. cit., supra, n. 46 at 170. Bessman and Swazey report that there have been at least "half a dozen malpractice suits involving PKU." op. cit., supra, n. 47 at 72.

genetic counseling. Some geneticists contend that

It is probably unjustified on ethical grounds to mount large-scale screening programs for disease or carrier detection in conditions where the patient and carriers cannot be offered specific effective medical therapeutic alternatives, including intrauterine diagnosis and abortion.<sup>189</sup>

This reference to abortion raises sensitive policy questions, including whether or not public funds should be used to provide abortion services. If society's resources are expended in order to provide families with information that is required for intelligent reproductive decision-making, can it then deny them the option to implement their decision, an abortion being one option they might choose?

Genetic counseling also has an important role to play in screening programs. First, it can provide couples with the basic genetic information required to make informed and intelligent decisions about subsequent pregnancies. And second, it provides the follow-up support needed to help those couples implement their decisions.

In providing information to couples, the genetic counselor will be able to explain the source and meaning of a particular defect and, after appropriate testing and analysis, inform them of the risk of its occurrence or recurrence. The lack of such information might lead to poor decision-making in either of two directions. Some couples might have additional children when the probability is high that their future offspring will be adversely affected. Or, conversely, couples with conditions in which the risk is very low may

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<sup>189</sup> Robert F. Murray, Jr., "Problems Behind the Promise: Ethical Issues in Mass Genetic Screening," The Hastings Center Report, Vol. 2, April 1972, p. 13.

have no further children as a consequence of unreasonable fear. A recent study of families with genetically-ill children found that

regardless of socio-economic class, birth order of the chronically ill child, religion, burden of care, and the hereditary nature of the condition, families continue to have children, whether purposefully or unplanned as do the parents of normal children. There is a strong indication, however, that this would not be the case if parents were aware of the risk involved in the transmission of genetic defects and if this information were coupled with knowledge of effective techniques to prevent pregnancy.<sup>190</sup>

Thus, the proper transmission of genetic information to couples might help them in planning their future families.

On a second level, counseling is needed to help couples adjust to and implement their reproductive decisions. A few examples drawn from case studies provide some insight into the various demands that would call for follow-up counseling. Fletcher studied the period of time following amniocentesis and found

the parents in considerable anxiety, and whatever problems existed in their marriage or family relationships were exacerbated . . . . If a marriage is troubled, the strains will most likely break forth in this period, testing to the limits the capacity of the couple to face their problem and make plans . . . . Counselors should be particularly attentive to the deeper personal problems which emerge in this period.<sup>191</sup>

Fletcher describes existing counseling centers as poorly set-up to deal with these problems, noting that perhaps "one in twenty-five centers would be sensitive to marital problems and fewer than that would have the means to help people."<sup>192</sup> He also found couples in "need for support and counseling

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<sup>190</sup> Harry Sultz, Edward Schlesinger and Joseph Feldman, "An Epidemiologic Justification for Genetic Counseling in Family Planning," The American Journal of Public Health, Vol. 62, November 1972, p. 1492.

<sup>191</sup> Fletcher, op. cit., supra, n. 134 at 56.

<sup>192</sup> Interview with John Fletcher, supra, n. 114.



at the time of therapeutic abortion and the deep depression suffered at the time."<sup>193</sup> Unfortunately, he notes that abortion counseling facilities are less than adequate for the task.

Counseling might also be required to follow up on the initial diagnostic tests. It is important to consider the total health needs of the patient and, to use the case of PKU, neglect of such needs regarding the dietary problems might result in more serious physical and emotional problems.

When treatment of a child involves restrictions in diet, it has broad implications for the entire family. How the parents feel about food, how much they use food as a weapon in the parent-child relationship, and how the other children in the home react, can mean the success or failure of the dietary regime. Consideration of the child as a member of a family that has many other responsibilities requires that medicinal and dietary care be obtainable without undue drain on family resources.<sup>194</sup>

There is need to assure, then, that any intervention into the genetic decision-making process will not be more injurious to the individual and/or his family than if such intervention had not occurred.

Counseling support might also help couples overcome the severe guilt feelings which often accompany the birth of an affected child. In the case of hemophilia, for example

there is a need to deal with the emotional upset that occurs - the shock at the discovery, the guilt and the self-blame which comes from the inevitable feeling that parents are somehow responsible for causing the disorder and the fears as to what hemophilia entails.

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<sup>193</sup> Fletcher, op. cit., supra, n. 134 at 51.

<sup>194</sup> Recommended Guidelines for PKU Programs for the Newborn, (U.S. Department of Health, Education and Welfare, Health Services and Mental Health Administration, 1971), Public Health Service Publication No. 2160, p. 9.

Helping the family through the upset is, of course, important in its own right, but it is also necessary for treatment purposes. Until the emotional problems are handled, it is very likely that the therapeutic efforts which require education of the parents about the reality of hemophilia will be hampered.<sup>195</sup>

Family follow-ups and counseling, therefore, might provide both the parents and the affected child with more effective genetic guidance.

A commitment to create such counseling services also entails the responsibility to ensure that the services reach prospective consumers. In his study of 250 counseling units, Sorenson found 25 per cent located in a hospital setting. He contends that

Hospital based medical genetics will probably increase significantly as the impact of the various intrauterine diagnostic procedures become more accepted . . . . Today, with various forms of heterozygosity detection possible, as well as amniocentesis, there is an increasing need for the delivery of medical genetics to be associated with the facilities of a hospital and laboratory.<sup>196</sup>

This setting, according to Sorenson, will not only permit the maintenance of adequate facilities, but will most likely increase "the proportion of lower

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<sup>195</sup> Lee Salk, M. Hilgartner and B. Granick, "The Psycho-Social Impact of Hemophilia on the Patient and His Family," Social Science and Medicine, Vol. 6, August 1972, p. 503. In the same article (p. 496), the investigators report that in "14 of the 25 cases, there appears to be a clear-cut deleterious impact, e.g. contributing to the breakup of the marriage or most leading to a psychological withdrawal by the husband from family relationships." For further evidence highlighting the presence and impact of parental guilt, see David G. Langsley, "Psychology of a Doomed Family," American Journal of Psychotherapy, 15:531-538, 1961; and Simon Olshansky, "Chronic Sorrow: A Response to Having a Mentally Defective Child," Social Casework, 43:190-193, April 1962.

<sup>196</sup> James A. Sorenson, "Sociological and Psychological Factors in Applied Human Genetics," Department of Sociology, Princeton University, October 1971, pp. 14 and 15.

socio-economic groups receiving counseling."<sup>197</sup>

Consideration should also be given to the nature of the delivery mechanism.

A prime requirement for any mechanism we devise to deliver service is that it be sufficiently flexible so that we can individualize the service package to fit in not only with abilities and peculiarities of the agents who are dispensing service but also with the individual differences of the consumers who receive them, and the communities and settings in which such services are provided and utilized.<sup>198</sup>

The importance of this point can be sufficiently demonstrated by the recent attempts of the National Institutes of Health to promote an employee sickle-cell testing campaign. The program was cancelled; one of the major reasons given for this was that it was "not organized by blacks with black feelings in mind."<sup>199</sup> The Institute of Society, Ethics and the Life Sciences has recommended that "From the outset program planners should involve the communities affected by screening in formulating program design and objectives, in administering the actual operations of the program, and in reviewing results."<sup>200</sup>

I. Screening Facilities and Organization: The provision of adequate and functional diagnostic facilities must also receive high priority from program planners. For maximum quality control and personnel expertise, testing should probably be done in regionally centralized laboratories so that costs can be minimized and the training of qualified personnel more easily accomplished.

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<sup>197</sup>Sorenson, *op. cit.*, *supra*, n. 169.

<sup>198</sup>Rudolph Hormuth, "Organization of Community Services in Phenylketonuria," in Anderson and Swaiman (eds.), *op. cit.*, *supra*, n. 46 at 165.

<sup>199</sup>Victor Cohn, "Black Health Care Lag Cited," *Washington Post*, November 15, 1972, p. A12.

<sup>200</sup>*Op. cit.*, *supra*, n. 154 at 1130.

Because "biochemical assays necessitate facilities that often are well beyond the means of the average clinical laboratory, [it] simply is not feasible for one laboratory to perform all of the tests now possible . . ."<sup>201</sup>

Thus,

A screening program should be carried out in conjunction with a facility large enough to handle a volume of samples sufficient to detect several positive cases per year to assure experience in laboratory diagnosis. Efficiency is materially increased when a single central laboratory is utilized. The development of a system of quality control on a statewide or regional basis should be considered to insure a high degree of reliability of results.<sup>202</sup>

Similar consequences to those which resulted from the impact of public pressure on the evolution of PKU legislation (supra, pp. 59-60) might also occur during management of diagnostic facilities and the provision of follow-up services. Past experience with establishing abortion facilities to meet expanding demands<sup>203</sup> and the difficulties which appear to be emerging in the

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<sup>201</sup> "Geneticists Press for Regionalization," Laboratory Management, Vol. 10, October 1972, p. 25.

<sup>202</sup> Recommended Guidelines for PKU Programs for the Newborn, op. cit., supra, n. 194 at 4. Such a network has been established by the National Genetics Foundation, Inc. Each of their 45 centers in the United States and Canada is staffed and equipped to perform the biochemical and chromosomal analyses necessary to diagnose the most common genetic diseases. In addition, some of the centers have the specialized personnel and facilities required for diagnosis of one or more rare genetic defects. All of the centers are staffed to provide genetic counseling and follow-up to any diagnosis. See their brochure "Genetic Counseling and Treatment Network," (National Genetics Foundation, Inc., 250 West 57th Street, New York 10019).

<sup>203</sup> Neubardt and Schulman discuss the problems which resulted in New York following the changes in its abortion law and conclude that "Abortion has exposed in rather vivid fashion the weaknesses of our medical institutions." op. cit., supra, n. 68 at 105-106.

development of hospital cardiac programs,<sup>204</sup> demonstrate the problems confronting program planners caught up in a cross-current of public pressure. The problems may already be emerging in the newly-enacted sickle-cell screening programs. Virginia, like a number of other states, hurried to join the bandwagon of states with sickle-cell testing programs. However, Virginia's Department of Health reports that "Sufficient funds have not been appropriated for recruiting or hiring the appropriate number of genetic counselors. . . . Until additional funds are appropriated, we are simply unable to meet all of the responsibilities placed upon us by the new legislation."<sup>205</sup> The consequences which result, and which merit emphasis, are not only that the necessary facilities and services will not be provided, but that a "confidence gap" is created between those designated to provide and perform the services and those who are to receive them. Such a situation obviously benefits no one and, in the final analysis, is probably counterproductive.

J. Screening Costs: There is also a need to consider the cost of screening and counseling services. The use of various diagnostic tests and extensive laboratory work can be quite expensive (for example, a typical examination with ultrasound of a potentially abnormal pregnancy can cost as

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<sup>204</sup>The Inter-Society Commission for Heart Disease Resources reported that hospitals are under new public pressure to enlarge their cardiac programs. The Commission warned that "With the introduction of new techniques for coronary-artery surgery hospitals are again being stimulated to expand their surgical programs and there is evidence we may again see a proliferation of poorly planned units with costly duplications of facilities and suboptimal care." See "Hospitals Warned on Heart Surgery," Washington Post, October 16, 1972, p. A15.

<sup>205</sup>Patricia Hunt, Director, Bureau of Child Health, in a letter to this author, October 6, 1972.

high as fifty dollars). Systematic study is needed of the costs involved in providing genetic services to the population and the extent to which such services are not used because of a family's financial circumstances. Also important is the role of private health insurance in helping families absorb the costs of genetic services. At this time,

No insurance company . . . recognizes the concept of preventive medicine. The fetus is not recognized as a patient. Cytogenetics is not, for the most part, recognized. The insurance situation seems particularly outrageous when one stops to consider that preventive medicine will be the mainstay of health care in the next century.<sup>206</sup>

As the availability and demand for genetic services increase, the question of costs will become an increasingly important matter.

K. Screening and Genetic Information: The collection and dispersion of genetic information acquired through screening programs also raises important policy questions. As more and more genetic information about individuals and their families is accumulated, how should it be used? What protection should be guaranteed to the individual to whom such information refers? Those who establish data-gathering systems need to be aware of the possible abuse of the information which they possess.

The management of screening programs carries with it two inherent potential sources of abuse. First, in a large-scale screening program

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<sup>206</sup> Carlo Valenti, quoted in Laboratory Management, Vol. 10, October 1972, p. 23. Geneticist Valenti reported that Blue Cross/Blue Shield has "agreed to partially cover the cost of diagnosis depending upon the type of policy held by the patient. However, the reimbursement schedule which they have offered is still less than adequate: \$25-40 for a chromosome analysis and \$15-25 for a buccal smear. These figures compare with hospital charges of \$100 for a diagnosis based upon leukocyte cultures, \$250 for a diagnosis based upon amniotic fibroblast cells, and \$40 for a buccal smear."

the responsibility for the quality and quantity of care that a patient receives rests with a team of medical experts rather than in the context of the traditional doctor-patient relationship. Thus, both the medical and ethical responsibilities to the patient are more diffused and consequently more difficult to fix. Experience with PKU screening illustrates this point.

Not only are records of tests filed in several different places in the state health department, but the entire preventive medicine apparatus of the state, including psychology, nursing, statistics, social services, nutrition, and education, is alerted to the condition. In this process, information which could seriously affect an individual for life is passed about among nonprofessional, nonmedical personnel who have no legal or moral responsibility to the individual.<sup>207</sup>

The second potential source of abuse arises from the use of computerized data-gathering techniques. A vast network of screening programs not only calls for the collection of large amounts of data, but also requires that such data be rapidly and efficiently stored, retrieved and transmitted between diagnostic centers. The ability of the computer to meet these demands makes it an ideal tool for such data management. The problems posed by the use of computers are not new. They simply change the economics and nature of processing information in ways that could result in the abuse of civil liberties. Questions regarding what data is to be collected, for what purposes, to whom it will be made available, and what mechanisms will exist for individuals to obtain and contest such data are all matters for public policy. A recent report of the National Research Council's Computer Science and Engineering Board describes the challenge to policy-makers.

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<sup>207</sup>Bessman and Swazey, op. cit., supra, n. 47 at 73.

Our task is to see what appropriate safeguards for the individual's rights to privacy, confidentiality, and due process are embedded in every major record system in the nation, particularly the computerizing systems that promise to be the setting for most important organizational uses of information affecting individuals in the coming decade.<sup>208</sup>

The risks involved in the collection of confidential information from any part of the population are accepted by society because of the presumed benefits of using this information. In the case of genetics, for example, the identification of heterozygous carriers would be of great value in estimating the gene frequency among different population. From this information one could calculate the number of individuals who are likely to be affected within the particular population. Thus, better planning for and control of genetic disease are potential benefits to be derived from screening programs. There is a need, therefore, to strike a balance between the community's requirement for information and its subsequent use and the individual's rights of privacy.

L. The Misuse of Information: In developing an appropriate information policy, one should bear in mind the kinds of abuses that might affect a "defective" individual. One such abuse is the possibility of encumbering him with a lifelong public stigma. A diagnostic medical label can destroy or distort relationships within a family and can close access to many of the normal channels and outlets usually open to people. Such influence can result in two ways: first, through affecting an individual's attitudes, his

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<sup>208</sup> Quoted in Gerald S. Schatz, "Computers and Privacy: Continuing Questions of Civil Liberties," News Report, Vol. 22, December 1972, p. 5. (News Report is a monthly publication of the National Academy of Sciences).



image of himself, his self-confidence and, therefore, his involvement in an activity; or second, by identifying a person in such a way so that he is systematically discriminated against. Past experience with genetic diseases illustrates this point clearly. Persons with Huntington's chorea, a disease characterized by progressive mental deterioration, will probably manifest signs of the disease by age 35, but may not have symptoms until a much later age. Until that time they are quite capable of functioning normally. However, the stigma which is often associated with a family with a history of the disease has resulted in "great secrecy within the family because of the fear of social, economic, or legal penalties should the knowledge be made public."<sup>209</sup>

The case of sickle-cell anemia illustrates the problems incurred by the innocent carrier of the disease. The major problem is confusing the person who has the disease with the person carrying the trait, but not the disease symptoms. A recent report expressed concern about the

dangers of societal misinterpretation of similar conditions and the possibility of widespread and undesirable labeling of individuals on a genetic basis. For instance, the lay public may incorrectly conclude that persons with sickle trait are seriously handicapped in their ability to function effectively in society . . . Extreme caution should therefore be exercised before steps that lend themselves to stigmatization are taken.<sup>210</sup>

A consequence of such stigmatization is that "much unnecessary anxiety on the part of parents and trait carriers and psychologic harm occurs when some

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<sup>209</sup> John Whittier, Audrey Heimler and Charles Korenyi, "The Psychiatrist and Huntington's Disease (Chorea)," American Journal of Psychiatry, Vol. 128, June 1972, p. 1550.

<sup>210</sup> Institute of Society, Ethics and the Life Sciences, op. cit., supra, n. 154 at 1132.

persons are identified as carriers of sickle-cell trait without an understanding of the harmlessness of their trait."<sup>211</sup> Individuals might come to be regarded as physically weaker or less fit. An example drawn from experience with another disease may help to illustrate the possible harm. Dr. Nicholas Hobbs, director of the staff for a five-agency federal study of labeling, has reported "growing evidence that for a child to be labeled anything - whether the label is 'mentally retarded' or 'gifted' - influences what social system he gets into and shapes his whole future."<sup>212</sup> Hobbs cites the example of a young child who was found to have a heart murmur. He was treated differently by his parents, "sheltered and not allowed to play with other children." Five years later doctors found the child's heart perfectly good, but by then "he had already developed a picture of himself as having heart disease and had taken on a restricted life-style that may never fully reverse." Great care must be taken, therefore, to avoid "overprotecting" carriers when it might later result in their adopting unnecessary and restrictive life-styles.

Another problem has been the denial to some sickle-trait carriers of employment opportunities or life and health insurance. For example there are reports that an airline stewardess was grounded after the airline discovered she carried the trait.<sup>213</sup> Also, insurance companies "have been

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<sup>211</sup> Beutler, et. al., op. cit., supra, n. 177 at 1486.

<sup>212</sup> Quoted in Suzanne Dean, "Study Probes Labeling of Children as Retarded," Washington Post, September 6, 1972, p. A8.

<sup>213</sup> Rudolph Jackson, Coordinator, National Institutes of Health sickle-cell disease program, quoted in B.J. Culliton, "Sickle Cell Anemia: The Route from Obscurity to Prominence," Science, Vol. 178, October 13, 1972, p. 141.

changing or raising the premiums or dropping insurance on persons with the trait . . ."<sup>214</sup> Thus, "sickle-cell testing has shown up in employment records, in insurance company records, and is becoming more and more abused by people who do not understand the nature of the disease."<sup>215</sup> While certain carriers of the sickle trait can experience some problems where the oxygen supply is diminished, most carriers will never have any problems and there is "no evidence that trait carriers have a higher risk of disease or a shorter than normal life-span."<sup>216</sup> This stigmatization of sickle-cell carriers has "created emotional resistance among many persons to sickle cell screening and genetic counseling . . ."<sup>217</sup> and consequently, the effectiveness of such programs has been greatly impaired.

The XYY chromosome abnormality presents yet another problem associated with stigmatization. This sex anomaly occurs in males with two Y chromosomes and one X chromosome (the normal chromosome complement for males is one of both X and Y chromosomes). The controversy which surrounds this aberration concerns the extent to which its presence predisposes an individual to engage in antisocial and violent behavior. A review of the literature indicates that the controversy is far from resolved. On the one hand, there are studies

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<sup>214</sup>Rudolph Jackson, quoted in "Bias Against Sickle Trait Victims Probed," Washington Post, November 14, 1972, p. A6. Some insurance firms have been reported to charge trait carriers as much as "150 per cent of the usual premium . . ." Joseph Christian, quoted in Culliton, op. cit., supra, n. 213 at 142.

<sup>215</sup>Mona Blake, School Board, Fairfax County, Virginia, quoted in "Sickle Cell Examination is Opposed," Washington Post, November 1, 1972, p. B9.

<sup>216</sup>Christian, op. cit., supra, n. 213.

<sup>217</sup>Cohn, op. cit., supra, n. 153 at A1.

which suggest a correlation between the XYY abnormality and certain types of aberrant behavior, with one concluding that "the additional Y-chromosome genetically predisposes the 47, XYY male to the development of a psychopathic personality and to consequent aberrant behaviors and antisocial conduct."<sup>218</sup>

There is equally persuasive evidence, however, which suggests that there is no strong correlation between the presence of the XYY chromosome complement and a particular type of behavior. Two researchers recently claimed that the suggestion that "XYY males are uncontrollably aggressive psychopaths appears to be nothing more than a myth promoted by the mass media."<sup>219</sup> In addition to the mixed findings suggested by these studies, there are also questions regarding the methodological and conceptual approaches employed in the investigations.<sup>220</sup> In light of this continuing debate, therefore, any attempt to develop policy which seeks to respond to the needs of individuals with the XYY anomaly would be premature.<sup>221</sup> The present state of knowledge does not

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<sup>218</sup> W.M. Court Brown, "Males with an XYY Sex Chromosome Complement," *Journal of Medical Genetics*, Vol. 5, 1968, pp. 348-49. Also see Lytt Gardner and Richard Neu, "Evidence Linking an Extra Y Chromosome to Sociopathic Behavior," *Archives of General Psychiatry*, 26: 220-222, March 1972.

<sup>219</sup> Seymour Kessler and Rudolf H. Moos, "XYY Chromosome: Premature Conclusions," *Science*, Vol. 165, August 1, 1969, p. 442. Also see S. Wiener and G. Sutherland, "A Normal XYY Man," *Lancet*, 2:1352, December 21, 1968.

<sup>220</sup> Saleem A. Shah (ed.), *Report on the XYY Chromosome Abnormality*, (National Institute of Mental Health: U.S. Government Printing Office, 1970), Public Health Service Publication No. 2103, pp. 23-27.

<sup>221</sup> The questions for policy consideration might include how much effort should be directed toward rehabilitating criminals if the underlying basis for their abnormal behavior is genetically determined. Or, how should the XYY individual be dealt with both prior to and following the commission of a crime? Since the XYY chromosomal abnormality can be detected in utero by amniocentesis, does society have a right to intervene into the reproductive decision of couples found to have conceived an XYY child?

permit any definitive statement regarding the possible link between the XYY complement and certain types of behavioral pathology. This lack of consensus highlights the danger "that incomplete or inadequate understanding of the phenomena might possibly become embedded into public policy or legislative enactments."<sup>222</sup> Thus, there is a need for more systematic data collection and research into this problem, and others like it, in order to provide the necessary information from which policy decisions can be made. While such research itself creates problems regarding the confidentiality of data and individual privacy, the problem of stigmatization is apparently an immediate one. A recent investigation concluded that XYY men had been falsely stigmatized<sup>223</sup> and it is not unreasonable to assume that such information, when improperly understood, might affect a man's opportunities for gaining employment or obtaining parole, or prejudice his judicial proceedings.<sup>224</sup> Furthermore,

presumptions that a person's chromosome pattern clearly disposes him toward aggressive and antisocial behavior could lead to further stigmatization of that individual. Responses from others interacting with him might be of

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<sup>222</sup> Saleem A. Shah, "Recent Developments in Human Genetics and Their Implications for Problems of Social Deviance," in Daniel Bergsma (ed.), Advances in Human Genetics and Their Impact on Society, (Birth Defects: Original Article Series, Vol. 8: The National Foundation - March of Dimes, July 1972), p. 79.

<sup>223</sup> G.R. Clark, M.A. Telfer, D. Bajer and M. Rosen, "Sex Chromosomes, Crime and Psychosis," American Journal of Psychiatry, 126: 1659-63, 1970.

<sup>224</sup> Recently, a public-interest group threatened a law suit regarding a study inquiring into the frequency of XYY males in a population of children. The group felt it an invasion of privacy to get information about an individual that might guide his future treatment. They took the view that the law might use this information in some way that would adversely affect the individual. For example, if it was known that an individual was an XYY, there might be a greater tendency to judge him guilty if arrested by police. See Robert Cooke, quoted in Maureen Harris (ed.), op. cit., supra, n. 105 at 82.

a form that would tend to promote aggressive behavior, thereby making a possible unwarranted assumption become a self-fulfilling prophecy.<sup>225</sup>

Care must be taken, therefore, to guard against such abuses.

M. Safeguarding Research Data: As suggested earlier, research into the XYY problem will create its own difficulties. It is important, then, that the requirements of rigorous scientific research be balanced with the proper respect for and protection of the rights and welfare of the subjects under study. The proper protection of the rights of research subjects requires policy that will safeguard confidential records and protect access to such information. Unfortunately, only eleven states have statutes that recognize the confidentiality of general research information of a public health nature.<sup>226</sup> Investigators thus face serious difficulties in protecting such information from court subpoenas. Even when such statutes are in operation, however, they are often "overly broad in regard to the possible range of material considered confidential within the statutes, and thus the researchers and even more importantly the subject may be misled in relying on a statute that might be given a narrow judicial construction."<sup>227</sup> It would seem to be an appropriate role of the legislature to formulate more discriminatory models for safeguarding the confidential nature of research data. The XYY anomaly, as well as experience with other genetic abnormalities, illustrates the potential problems of data management in large-scale screening programs.

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<sup>225</sup> Shah (ed.), op. cit., supra, n. 220 at 9.

<sup>226</sup> Ralph K. Schwitzgebel, "Confidentiality of Research Information in Public Health Studies," Harvard Legal Commentary, Vol. 6, 1969, p. 192.

<sup>227</sup> Ibid., p. 196.

N. Public Education: Much of the stigmatization cited above can be lessened, and perhaps to a great extent avoided, if the public can be educated about the nature and consequences of genetic disease. In the case of sickle-cell disease, the issue is not merely identifying trait carriers, but giving those individuals and society better information about what being a trait carrier means. An educated public can thus be a means of "reducing the potential risk that those identified as genetically variant will be stigmatized or ostracized socially."<sup>228</sup> So far, efforts in this direction have not been very successful. The excessive pessimism and hostility among blacks toward genetic screening programs has been attributed to the "large amounts of 'unfortunate sensationalism' and badly informed 'scare campaigns' in TV and newspapers."<sup>229</sup> Clearly, a more carefully constructed and broad-based educational campaign should accompany genetic screening.

Education, however, has other important functions to perform. Studies indicate that most people are unaware of the opportunities for genetic services, with persons of the lower socio-economic classes relying "primarily on family and friends for information, [which] means not only that they are not likely to be as informed as others, but that there is an increased chance that they will in fact receive incomplete and often erroneous health information."<sup>230</sup> Thus, an important task will be to make people aware of available medical opportunities. Education is also necessary if persons are to be able to make

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<sup>228</sup> Institute of Society, Ethics, and the Life Sciences, op. cit., supra, n. 154 at 1130.

<sup>229</sup> Cohn, op. cit., supra, n. 153.

<sup>230</sup> Sorenson, op. cit., supra, n. 196 at 7-8.

intelligent decisions regarding their future medical and genetic status. A survey of PKU parents demonstrates the educational challenge ahead. The survey was designed to find out how much such parents knew about their circumstances; the results were not very encouraging.

1. 61% did not know the disorder was inherited;
2. 58% did not understand the importance of early diagnosis;
3. 56% said that they had never discussed the condition with a professional source;
4. 56% did not know that the condition can be treated with a special diet.<sup>231</sup>

If genetic screening programs are to be effective in ameliorating the effects of genetic disease, an educated public is essential.

#### IV. Genetic Counseling

Genetic counseling is one of the most important means for transforming the results of medical and genetic's research into measures designed to provide immediate and practical aid to individuals. The emphasis of the following discussion will be on the training requirements for genetic counseling, the possible roles that genetic counselors might assume in performing their counseling services, doctor-patient communication, and the responsibilities of the genetic counselor to his patients and society, particularly with respect to the information to which he has access.

A. Training Requirements: Today there are about 200 genetic counseling units in the United States. With the increasing awareness of the need for counseling, the number of these units should proliferate. The services offered by the units, however, will only be as good as the counseling personnel which

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<sup>231</sup> Lloyd Kramer and Benjamin White, "A Survey of Families and Relatives of Proven Phenylketonuria (PKU) Patients in Maryland," Paper presented at 92nd Annual Meeting, American Public Health Association, 1964.



provide them.

What appears to be developing . . . is a rapidly expanding knowledge base permitting increasingly refined prediction and control of genetic and chromosomal problems, but no concomitant professional or organizational locus of training, socialization and control. What this means is that genetic counseling as currently practiced exhibits considerable diversity. With no singular professional training experience, counselors rely largely on their individual medical backgrounds, local institutional constraints, as well as the specific demands placed on them in the counseling session to shape their counsel.<sup>232</sup>

As a result of this diverse and often narrow educational and training experience, two problems emerge. First,

because most physicians lack adequate training in diagnosing genetic defects, misdiagnosis and inappropriate advice can be serious problems. For example, if a couple are told that a given problem is genetic and accordingly opt for sterilization, they have taken an irreversible step. If the doctor was wrong, not only has he caused the couple much grief, but he is legally liable. The current structure of genetic counseling facilities combined with the lack of diagnostic capacity make such problems likely. In addition, given the current lack of training in medical genetics, practicing physicians probably ignore the genetic aspects of many diseases.<sup>233</sup>

The second problem which stems from this diversity in backgrounds is that

Professional counselors . . . tend to erect fences around their area of counseling interest and, by fiat, allow other professionals to give genetic facts but not counseling.

Counseling preserves established by vested areas of interest also increase the likelihood that families will miss vital pieces of information. When no one person carries

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<sup>232</sup>James R. Sorenson, "Factors Shaping Decision Making in Applied Human Genetics: Professional and Client Perspectives," Department of Sociology, Princeton University, August 1972, p. 4.

<sup>233</sup>Sorenson, op. cit., supra, n. 169 at 27.

the primary responsibility for organizing the genetic information and counseling the family, important aspects of information may be overlooked.<sup>234</sup>

There is a need, therefore, to define qualifications and to provide proper training for genetic counselors and to consider appropriate guidelines for the conduct of counseling services. Ladimer suggests that appropriate standards should cover the

- (1) definition of the field or process of genetic counseling;
- (2) scope of services; (3) practitioners qualified to serve;
- (4) institutional and other settings suitable for counseling;
- (5) protection of interests; (6) relation to other fields, professions and services; (7) methods for evaluation; and
- (8) professional and community obligations.<sup>235</sup>

Certainly, an important goal of training counselors should be to sensitize them to the wide variety of needs and expectations that may be expressed by their patients.

B. Role Orientation: Sensitizing the genetic counselor to patient needs and expectations naturally raises the question of how he should relate to his patients. The role orientation of counselors has been a subject of considerable discussion. Two basic positions can be distinguished. On the one hand, there are those who view the counselor as an informer, whose task is simply to inform the couples of the risks involved. They see any attempt on the counselor's part to influence the decisions of those whom he counsels as beyond his professional responsibility as a counselor. On the other hand, there are those who view the counselor as a wise advisor, one whose concern for the patient exceeds a mere presentation of the facts and calls for a closer

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<sup>234</sup> Reisman and Matheny, op. cit., supra, n. 15 at 27.

<sup>235</sup> Op. cit., supra, n. 186 at 13.

involvement with his counselees. No matter which of the two views one adopts, it is important to recognize the influential role which a counselor can assume. Sorenson has observed that

counselees are often informationally dependent on the counselor for not only a technical diagnosis and assessment of their situation, but they seek in addition some assistance in giving meaning to the condition they find themselves in, a condition of calculated risk, but a condition for which there are few behavioral precedents as to how to interpret these risks or how to make sense out of them.<sup>236</sup>

Thus, lacking adequate and meaningful information, a couple seeking help is confused and worried. They are searching for someone with authority to answer their questions and this "dependency role" may make them more susceptible to the counselor's own views. And it is difficult, if not impossible, for the feelings of the counselor not to be conveyed to his

patients.<sup>237</sup> The opportunities which exist for counselors to influence the decision of a couple are illustrated by the following example.

If the couple is facing the risk for an autosomal recessive disorder the counselor can tell the couple that they have a three in four chance of having a normal child. He might do this if he thinks that the couple ought to have more children. On the other hand, if he is pessimistic and believes that the couple ought not to chance reproducing he might say that they face a risk of one in four that the child will be abnormal. In both cases the same factual

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<sup>236</sup> Op. cit., supra, n. 232 at 15. Sorenson presents an excellent description and analysis of the context in which genetic counseling occurs, emphasizing the evolution of a new doctor-patient relationship and its implications for applied genetic decision-making.

<sup>237</sup> Fletcher's study tends to support this point, with the finding that "The counselor's wishes for outcomes in a case will be conveyed directly or indirectly to the patient." (emphasis added) Op. cit., supra, n. 134 at 60. The importance of recognizing and analyzing the nature of this influence is stressed by Sorenson, who writes that "the ultimate role of who makes final decisions regarding the use of genetic knowledge is usually less ethically and morally neutral than is the situation in the delivery of more standard medical services." Op. cit., supra, n. 196 at 19.

information is conveyed to the clients. In the first situation the counselor stresses normality, while in the second he stresses the potential abnormality. This variation is certain to have an impact on the decision of the clients.<sup>238</sup>

Thus, the genetic counselor's own biases may well become important factors influencing a family's decision.

C. Doctor-Patient Communication: The ultimate quality of genetic counseling will, to a large extent, depend on the interaction between the patient and the counselor. Of the various factors which contribute to this interaction, certainly one of the most crucial is the communicative process between the counselor and his counselees. How well do patients receive, comprehend, and apply the information given to them by their physicians? To what extent are instances of misunderstanding and distortion due to the patient's or physician's inability to "communicate"? Answers to these questions are crucial, for it may well be that shortcomings in the treatment of a chronic illness can be related to such misunderstanding or distortion.<sup>239</sup>

There appear to be at least three pertinent clearly-defined variables in the doctor-patient communicative process. First, there is the ability of the physician-counselor to communicate information to his counselees. Of what value is it to have pertinent information unless the counselor is able

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<sup>238</sup> Sorenson, op. cit., supra, n. 196 at 22.

<sup>239</sup> Undoubtedly, many individuals distort, forget, or reject the genetic information conveyed to them by the genetic counselor. A study of parental understanding of phenylketonuria concludes: "If exposing parents to medical information aims at improving their understanding of the illness or at favorably influencing the course of the child's illness, the present study provides no support for either contention," Maarten S. Sibinga and C. Jack Friedman, "Complexities of Parental Understanding of Phenylketonuria," Pediatrics, Vol. 46, August 1971, p. 222. The study's sample population included 42 families of children with PKU.

to deliver it effectively? But while it is the counselor's role to promote effective communication, it is a role, according to some, "for which most physicians have unfortunately had little training."<sup>240</sup> The problem resulting from poor communication is described by a recent study of doctor-patient communication in a pediatric clinic of a large hospital. The study found that physicians tend to be overly technical in the language they use with their patients. "In more than half of the cases we recorded the physicians resorted to medical jargon. This did not necessarily leave the patient dissatisfied; . . . It did, however, leave most of the mothers unenlightened about the nature of the child's illness."<sup>241</sup> This problem is also applicable to genetics. Unless the information is properly explained and understood, it may evoke unreasonable fear on the part of families. For example, in a follow-up study at a genetic counseling clinic, it was found that in some instances odds had no meaning to couples. "The mother of a child with a myelocoele remembered that she had been given a 1 in 25 risk, but said that if she had another child 'either it would or would not be affected and so the risk is 50/50'."<sup>242</sup> On the basis of her reasoning, this woman had adopted three children. Unquestionably, then, "attention to effective communication, a skill that should not be too difficult for any trained person to master, could make a valuable contribution to the quality of health care and its availability to the general population."<sup>243</sup> Genetic counseling would appear

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<sup>240</sup> Reisman and Matheny, op. cit., supra, n. 15 at 30.

<sup>241</sup> Barbara Korsch and Vida Megrete, "Doctor-Patient Communication," Scientific American, Vol. 227, August 1972, pp. 71-72.

<sup>242</sup> Carter, et. al., op. cit., supra, n. 163 at 282.

<sup>243</sup> Korsch and Megrete, op. cit., supra, n. 241 at 74.

to have much to gain from a concerted effort in this direction.

In a recent study in a congenital heart clinic with a well trained genetic counseling unit, it was found that after receiving genetic information only about 25 per cent of the families retained and understood the attendant recurrence risks.<sup>244</sup> Thus, even with highly-skilled genetic counselors, the reception and understanding of genetic information was significantly impaired. This leads to a second variable in the doctor-patient communicative process: the basic knowledge of biology and genetics that patients bring to the counseling session. Leonard and his colleagues found that "the substratum of biologic knowledge possessed by many parents is inadequate to support the information imposed upon it by the counselor."<sup>245</sup> In the long run, therefore, there is a need for a better-educated public. Of more immediate concern, however, is the need for systematic and empirical investigation into how counseling information is received and applied. Perhaps such information should be repeated. If so, how often and at what intervals? It might also be helpful to modify counseling services to the specific educational and socio-economic backgrounds of the consumers. How this might be most effectively and efficiently accomplished will require additional study.

While greater education is a necessary prerequisite for more effective doctor-patient communication, it is apparently not always sufficient for producing the desired effect, e.g. the family's understanding of the counseling information. In a study of PKU families, "Parents with greater education were

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<sup>244</sup> J.A. Reiss and V.D. Menashe, "Genetic Counseling and Congenital Heart Disease," Journal of Pediatrics, 80:655-656, April 1972.

<sup>245</sup> Leonard, et. al., op. cit., supra, n. 165 at 438.

no less inaccurate or distortion prone than those with less education . . . ." <sup>246</sup>

The investigators suggest that "the capability to understand illness might be considered an emotional phenomenon." <sup>247</sup> Thus, a third important variable is the context in which genetic counseling is provided. A recent study of genetic counseling cited "emotional conflict" as an inhibiting influence on a family's understanding of counseling information. <sup>248</sup> To what extent, then, do parents remember information given to them when the context is so emotionally charged? Is reinforcement required? If so, what forms should it take? Clearly, the emotional context of genetic counseling requires careful assessment when considering ways to improve the counseling process.

Doctor-patient communication, then, is an essential element of the counseling process and thus becomes an important criteria for designing and evaluating genetic screening programs. The three variables discussed above must be viewed as essential elements of doctor-patient communication, which, if carefully studied, evaluated and improved upon, could contribute to more effective control of genetic disease.

D. "Responsible" Genetic Counseling: How one defines the responsibility of the genetic counselor to his patients will depend, to a large extent, on the way one characterizes the practice of genetic medicine. There are those who contend that genetic screening and counseling are altering the paradigm of the

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<sup>246</sup> Sibinga and Friedman, op. cit., supra, n. 239.

<sup>247</sup> Ibid.

<sup>248</sup> Leonard, et. al., op. cit., supra, n. 165. The investigators noted (p. 435) that five of the families interviewed "observed that the genetic information given at the time of diagnosis or shortly thereafter was not retained because of emotional shock." Also, see supra, pp. 63-64.

traditional doctor-patient relationship and thus changing the nature of the physician's responsibilities within that context. Traditionally, the practice of medicine was primarily devoted to individual therapy, with the patient the responsibility of a single physician. Large-scale screening programs, however, have shifted the focus of attention from the individual to a larger population and from a single physician to team care. While medical codes regarding the professional responsibilities of the physician to his patient have been adequate for those problems arising from the traditional practice of medicine, new genetic technology and the kinds of medicine it makes possible may require a reevaluation of the ethical norms governing medicine. Perhaps the best illustration of the problems which can arise concerns the kinds of information that should be given to the patient.

Under the traditional doctor-patient relationship, the physician examines his patient and, on the basis of his diagnosis, then acts to prescribe the most effective alternative for alleviating the illness. In this arrangement, the patient assumes that the physician possesses superior knowledge concerning questions of medicine and health. This is not the case in genetic counseling. "There is no assurance that a counselor has any more expertise than the counselee in evaluating risks for recurrence of a problem or in estimating the ability of the family to adequately handle a problem, should it occur."<sup>249</sup> Under these circumstances, then, the counselor gives a couple information so that they can act, rather than as a prerequisite to his acting on them. Questions arise, however, concerning the status of a

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<sup>249</sup> Sorenson, op. cit., supra, n. 232 at 9.



physician's therapeutic privilege in the counseling context. What information should he give to his patients? Unfortunately, traditional ethical precepts offer little guidance. Consequently,

Since there are few normative guidelines outlining the information that should be given in the counseling sessions, other than the provision of minimal information about the disease and its recurrence risk, the actual information that is exchanged, and the degree to which this constitutes counsel, advice, or behavioral suggestion, varies depending on the particular conditions.<sup>250</sup>

The problems which may emerge from this unsettled situation can be demonstrated by the following "cases."

Earlier discussion of the XYY chromosome abnormality emphasized the inconclusive nature of research concerning its consequences on human behavior (supra, pp. 73-74). Assume for the moment that amniocentesis is performed on an expectant mother concerned that her child might be a Tay-Sachs baby. While no evidence of Tay-Sachs disease is found, the abnormal XYY chromosome abnormality is discovered. What should the counselor tell the mother? One question which this example raises is whether a counselor can simply act as an "informer," responsible only for providing his patients with the facts? In the case of the XYY anomaly, what are the "facts"? What consideration should the counselor give to the effect on the parents and their family situation if given this information? Might parental concern about the possible presence of an abnormality adversely influence their care of the child? A recent report on the XYY anomaly suggested that "parental expectations and apprehensions about possible - but as yet unknown or even non-existent -

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<sup>250</sup>Ibid., pp. 16-17.

problems, may well create certain difficulties and lead unwittingly to self-fulfilling prophecies."<sup>251</sup> Perhaps more importantly, should individual counselors, each with their own built-in biases and operating within the context of varied family situations, be given the responsibility to make such decisions? By whom and by what criteria should such responsibility be allocated? And if a child or young adult, while participating in a screening program to detect other sex anomalies is found to be an "XYY" should he be informed of this condition? Ramsey raises the question of "whether the individual might be endangered by the acquisition, in any society, of complete knowledge [or in the XYY case, of partial and as yet unconfirmed knowledge] of his behavioral genetics? Such knowledge may be too heavy for many to bear and still remain spontaneous and free in their personal lives."<sup>252</sup> Without any common ethical perspective, answers to these questions would undoubtedly vary from counselor to counselor and according to the situational context in which they occur.

In cases in which early detection of a disease cannot be accompanied by appropriate treatment for the patient or his family, the question arises whether the uncovering of the disease does more harm than good. The urgency of this question is demonstrated by the possible development of a safe and accurate test for presymptomatic detection of Huntington's chorea.<sup>253</sup> A

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<sup>251</sup> Shah (ed.), op. cit., supra, n. 220 at 26.

<sup>252</sup> Op. cit., supra, n. 86 at 174.

<sup>253</sup> H.C. Klawans, G.W. Paulson and A. Barbeau, "Predictive Test for Huntington's Chorea," Lancet, 2: 1185-86, December 5, 1970. The authors report using "levodopa" as their testing agent. They stress the need, however, for additional experimental testing and caution against hasty interpretations

reliable and accurate test will mean that persons who lack the deleterious gene will be reassured that the disease will not develop, and, thus, they will also be reassured that any children that they might have will be unaffected. But for those whose tests are positive, they will be confronted with the fact that their future will include gradual physical and mental degeneration. This possibility has led some to argue that "it is not unreasonable to withhold the use of a test of this sort until we have something tangible to offer to those who give a positive result," suggesting that "depression and the risk of suicide would be more or less inevitable."<sup>254</sup>

There are those who would object to this alternative, finding "no reason to deprive the patients involved of the right of decision to learn, early or late, their inevitable fate."<sup>255</sup> (emphasis added) It is important to remember that if the test is to be of value, persons with predictive signs of Huntington's chorea must refrain from having their own children. If such persons are to be informed, then it is imperative that the counselor carefully evaluate the emotional state of the patient prior to telling him. "Should the diagnosis be confirmed without proper preparation, serious behavioral or mood disorders may ensue, including suicide."<sup>256</sup> Thus, what is to be told and how it is to

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of test results, noting that "A positive result does not prove Huntington's chorea, it only increases the prediction coefficient . . . A negative result is still meaningless and requires new evaluation in years to come."

<sup>254</sup>David L. Stevens, "Test for Huntington's Chorea (cont.)," The New England Journal of Medicine, Vol. 285, August 12, 1971, p. 414.

<sup>255</sup>Willard Gaylin, "Genetic Screening: The Ethics of Knowing," The New England Journal of Medicine, Vol. 286, June 22, 1972, p. 1362.

<sup>256</sup>Whittier, et. al., op. cit., supra, n. 209 at 1550.

be told assume new proportions in the information equation established between the counselor and his patient.

While the above examples are single, isolated cases, they help to demonstrate some of the issues which may emerge concerning the counselor's responsibility in providing information to his counselees. A general overview of this responsibility highlights four additional policy questions. First is the question of whether persons are deprived of their freedom of choice when pertinent information is withheld. Without the knowledge necessary for making intelligent decisions, is the power to decide still meaningful? When arbitrating the question of what to communicate to the patient, it should be remembered that "For parents, genetic counseling can constitute a fundamental crisis, or emergency, in their reproductive careers. At issue is the decision as to whether to keep open or to close the social family biologically."<sup>257</sup> To what extent, then, should a "third party" be permitted to take that decision (in any meaningful sense of the term) away from a couple?

A second issue concerns the validity of the assumption that the withholding of information would be in the best interests of the patient. Some geneticists express the opinion that, in the case where there is no effective therapy for an illness, informing the patient and his family of his condition will do more harm than good. Knowledge of the condition

prior to its clinical manifestation may merely provoke increased patient or parental anxiety without offering them any positive reassurance. There will be little benefit to the patient and, for a time, at least, some possible degree of harm to the parents and patient, depending upon their emotional stability.<sup>258</sup>

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<sup>257</sup>Sorenson, op. cit., supra, n. 232 at 10.

<sup>258</sup>Murray, op. cit., supra, n. 189 at 10.

It is certainly questionable, however, whether the counselor will be able to determine what the "best interests" of his counselee are. Genetic counselors, unlike the family physician, are not well-acquainted with their patients and their families. Thus, some believe that "When counseling becomes much more routine, part of the accepted practice should not be the routine of withholding information from the counselees on the spurious grounds that the counselors know what is best for patients they hardly know at all."<sup>259</sup> There is also the problem of a physician's own values, which may differ from those of his patients, and the effect that they might have on his judgment to discern his patient's best interests. "The potential for conflict is especially great in genetic counseling in which the options elected depend on one's opinions about such controversial matters as the importance of the traditional concept of family, the morality of divorce and of abortion, . . ."<sup>260</sup>

The cornerstone of the doctor-patient relationship is the patient's trust in the integrity and ability of his physician. This poses a third question: If information that is withheld today is discovered later, what will be the effect on the relationship between the medical profession, and particularly genetic counseling, and its patients? Might there be a general loss of confidence in the medical profession brought about by the routine withholding of information? When could a patient be sure that he was being told all? The possible damage to the practice of medicine and its consequent impact on

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<sup>259</sup> Alexander M. Capron, "Ethical and Legal Aspects of Genetic Counseling," Paper presented at the First Advanced Symposium: Genetic Counseling, The New England Institute, Ridgefield, Connecticut, July 6, 1972, p. 4.

<sup>260</sup> Ibid., p. 5.

the health of the population must be added to the growing list of policy considerations.

The fourth and final broad policy question concerns the nature of the counselor's responsibility beyond his individual patient. Specifically, what is the responsibility of the counselor toward a patient's family and society? With respect to access to information, it has been suggested that "As a general rule all unambiguous diagnostic results should be made available to the person, his legal representative, or a physician authorized by him."<sup>261</sup> A question arises as to whether a patient's family should also be told, since this might give them the opportunity to evaluate intelligently their own health status. It has been suggested that the traditional, confidential doctor-patient relationship might be less important than people's "right to know about the risks that they run, whether infectious, toxic, or genetic."<sup>262</sup> And if such information is withheld, could the physician or screening program administrators be found legally negligent?

There may be instances in genetic medicine where the needs of the individual and those of society conflict. Considering the possible dangers of the presence of the XYY chromosome abnormality, does the counselor have the responsibility to forewarn the community and perhaps expose the XYY individual and his family to an undercurrent of social and legal pressures? And upon making a diagnosis of Down's syndrome and advising his counselees, to what extent should the counselor consider the costs to society of providing

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<sup>261</sup> Institute of Society, Ethics and the Life Sciences, op. cit., supra, n. 154 at 1131.

<sup>262</sup> John Littlefield, quote in Time, June 26, 1972, p. 51.

institutional care for that child? This issue requires considerable thought and leads one to ask if the individual physician is in a position to measure and evaluate the cumulative, and sometimes remote, effects of his individual acts? Should such considerations influence the case of his patient? It might well be that "The individual physician is unfaithful to the trust the patient places in him if he withholds a specific therapeutic agent in anticipation of some eventual perturbation of human ecology. Society, therefore, cannot possibly delegate such decisions to each physician."<sup>263</sup>

But how society and its institutions is to make these decisions is far from clear.

Genetic technology is becoming an increasingly important part of society's vast medical arsenal. Applying such knowledge, however, may create a myriad of problems. In the hope of stimulating discussion and focusing attention on the most pressing policy issues related to genetic technology, this paper has sought to identify and analyze some of the major problem areas. It is apparent that society must begin to make some conscious decisions regarding the use of this technology. How these decisions are made will affect not only the health of this generation, but that of many generations to come. These, then, are the promises and problems of genetic technology.

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Edmund D. Pellegrino, "Physician, Patients, and Society: Some New Tensions in Medical Ethics," in E. Mendelsohn, et. al., op. cit., supra, n. 47 at 80.

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